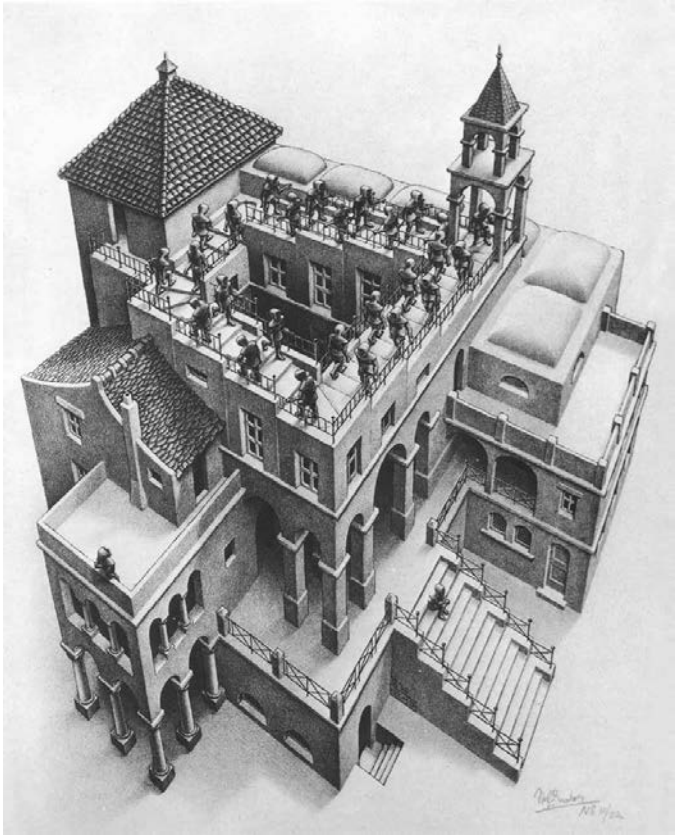
The child first and always

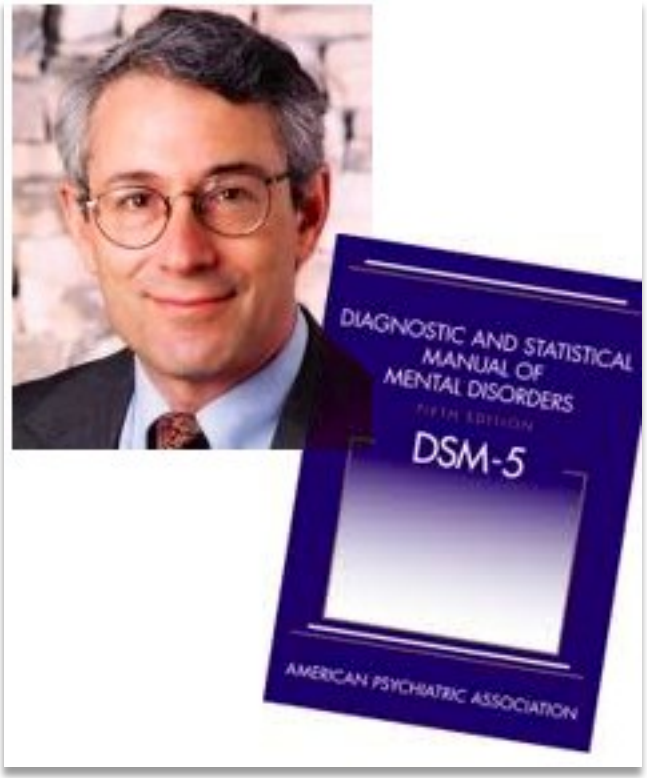
UCL

Phenotypic conundrums in the diagnostic appraisal of autism spectrum disorders




DSM-5
COLLECTION
Just Published!
DIAGNOSTIC AND STATISTICAL MANUAL OF MENTAL DISORDERS
DSM-5
AMERICAN PSYCHIATRIC ASSOCIATION
Pocket Guide
DSM-5
DESK REFERENCE
DIAGNOSTIC CRITERIA
DSM-5
American Psychiatric Publishing


DSM-5



Tom Insel
Director, NIMH
29th April 2013

The weakness is its lack of validity. Unlike our definitions of ischemic heart disease, lymphoma, or AIDS, the DSM diagnoses are based on a consensus about clusters of clinical symptoms, not any objective laboratory measure

“Patients with mental disorders deserve better”.

NIMH has launched the Research Domain Criteria (RDoC) project to transform diagnosis by incorporating genetics, imaging, cognitive science, and other levels of information to lay the foundation for a new classification system.

A diagnostic approach based on the biology as well as the symptoms **must not be constrained** by the current DSM categories,

Mental disorders are biological disorders involving brain circuits that implicate specific domains of cognition, emotion, or behavior

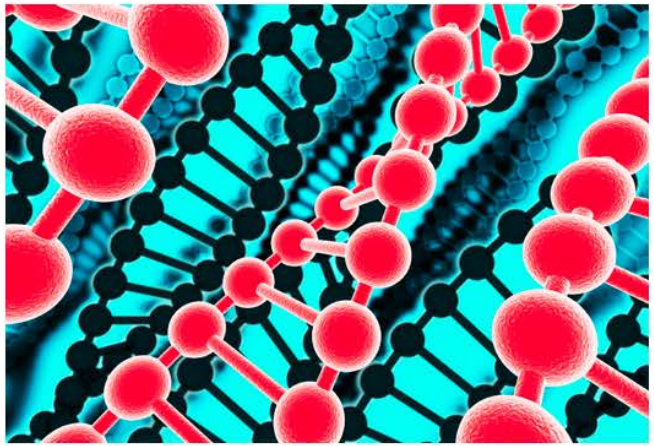
Each level of analysis needs to be understood across a dimension of function,

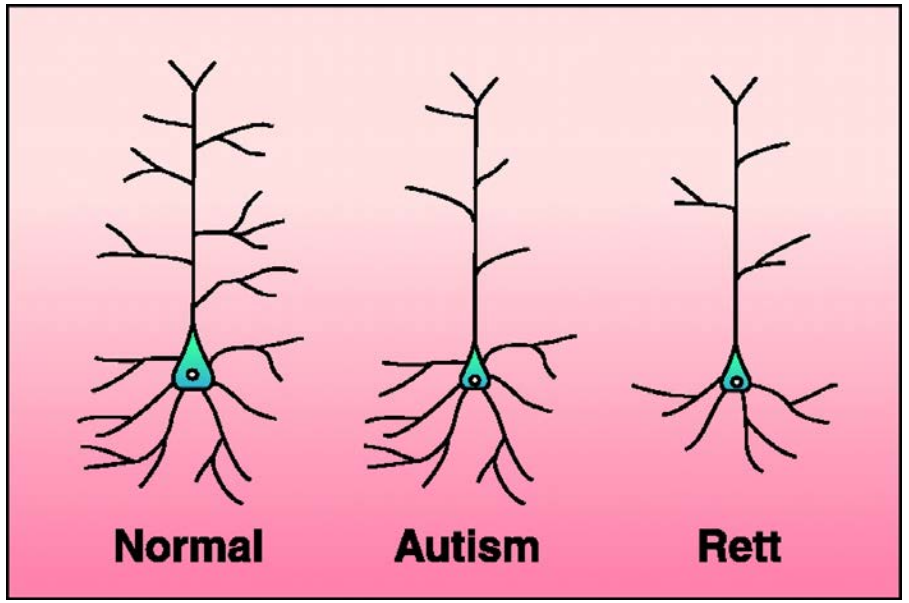
Mapping the cognitive, circuit, and genetic aspects of mental disorders will yield new and better targets for treatment.

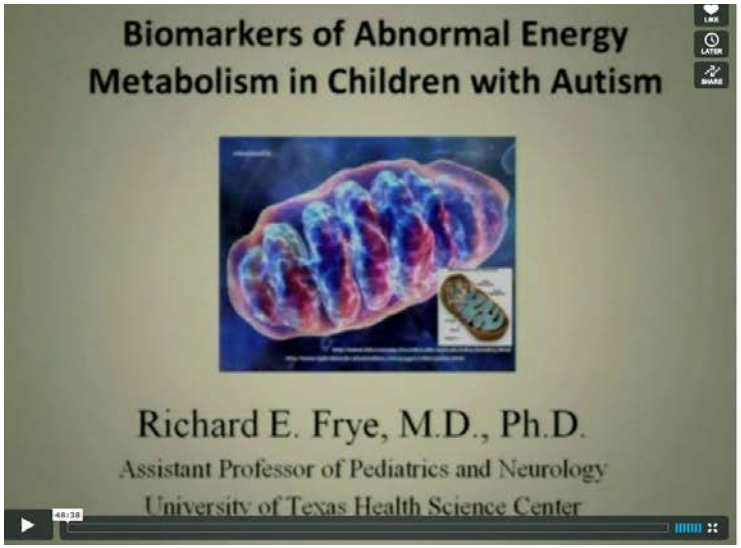
No biomarkers

Mouse mirrors severe form of autism

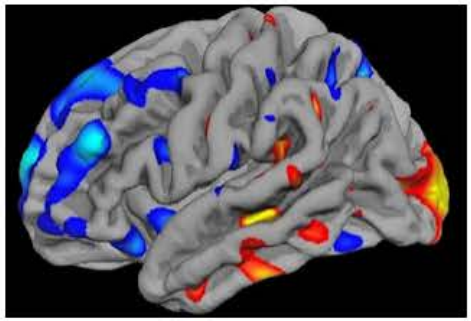
U. BUFFALO (US) — The first transgenic mouse model of a rare and severe type of autism is expected to improve understanding of the disorder and help researchers design more targeted treatments.







Autism Earthquake? The MRI Adult Autism Brain Scan Biomarker



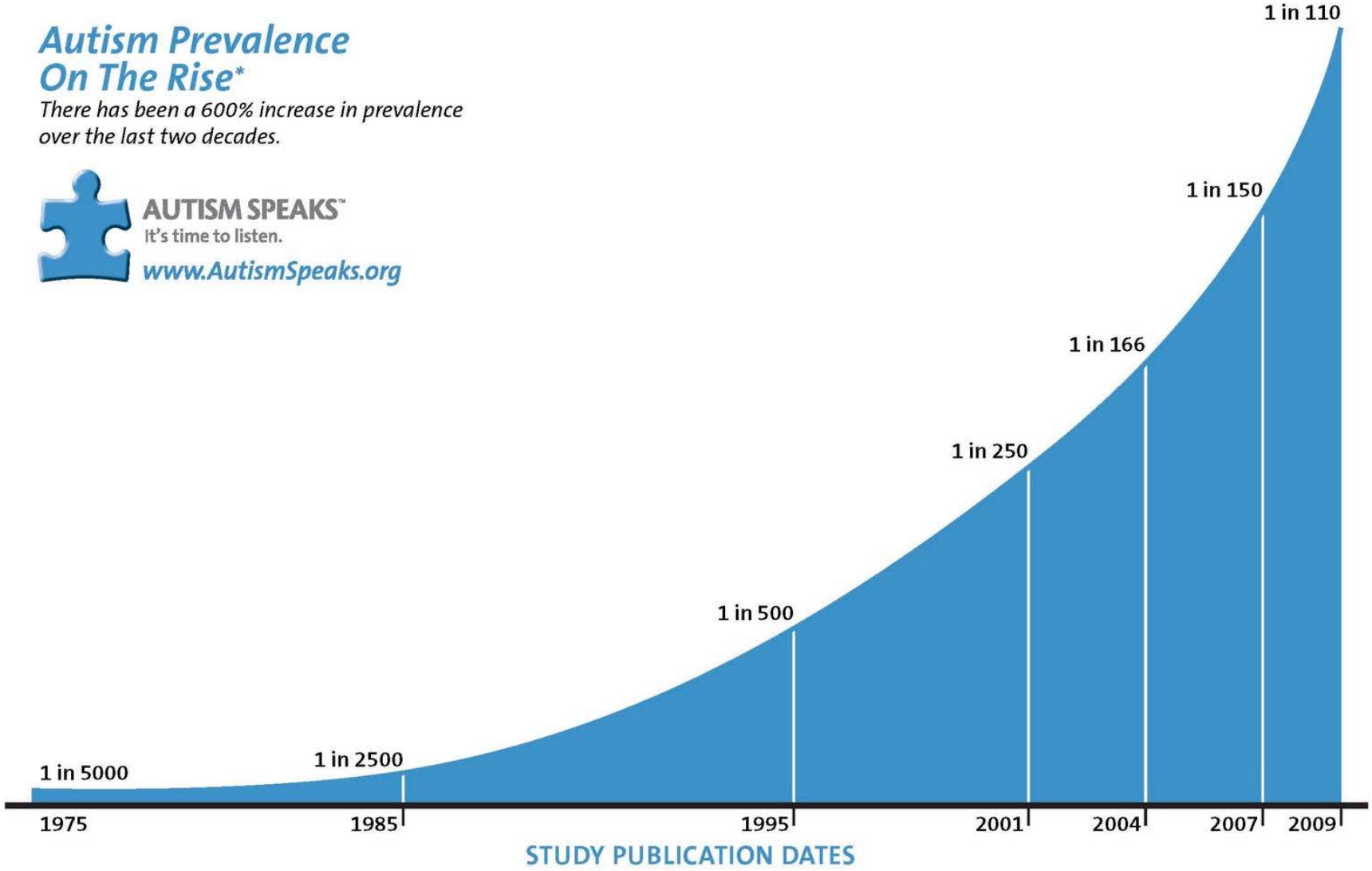
Every day brings reports of new autism studies, most of which are of little or no obvious value, studies which show that a group of high functioning autism subjects are very intelligent, studies which show that some families of autistic children are relatively affluent, studies which purport to define an autistic "smile" and so on.

News of a study which claims to have established, using MRI bran scans, a biomarker for autism disorders is different. An autism biomarker may have profound and immediate impact on the very definition of autism as "Socrates" of the New Republic has already mentioned in ***Autism Diagnosis by MRI Brain Scan***. Socrates gets right to the point while introducing a quote by one of the study authors Declan Murphy of King's College London:

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.

National Health Statistics Reports

Number 65 ■ March 20, 2013

Changes in Prevalence of Parent-reported Autism Spectrum Disorder in School-aged U.S. Children: 2007 to 2011–2012

by Stephen J. Blumberg, Ph.D., Matthew D. Bramlett, Ph.D., National Center for Health Statistics;
Michael D. Kogan, Ph.D., Maternal and Child Health Bureau;
Laura A. Schieve, Ph.D., National Center on Birth Defects and Developmental Disabilities;
Jessica R. Jones, M.P.H., and Michael C. Lu, M.D., M.P.H., Maternal and Child Health Bureau

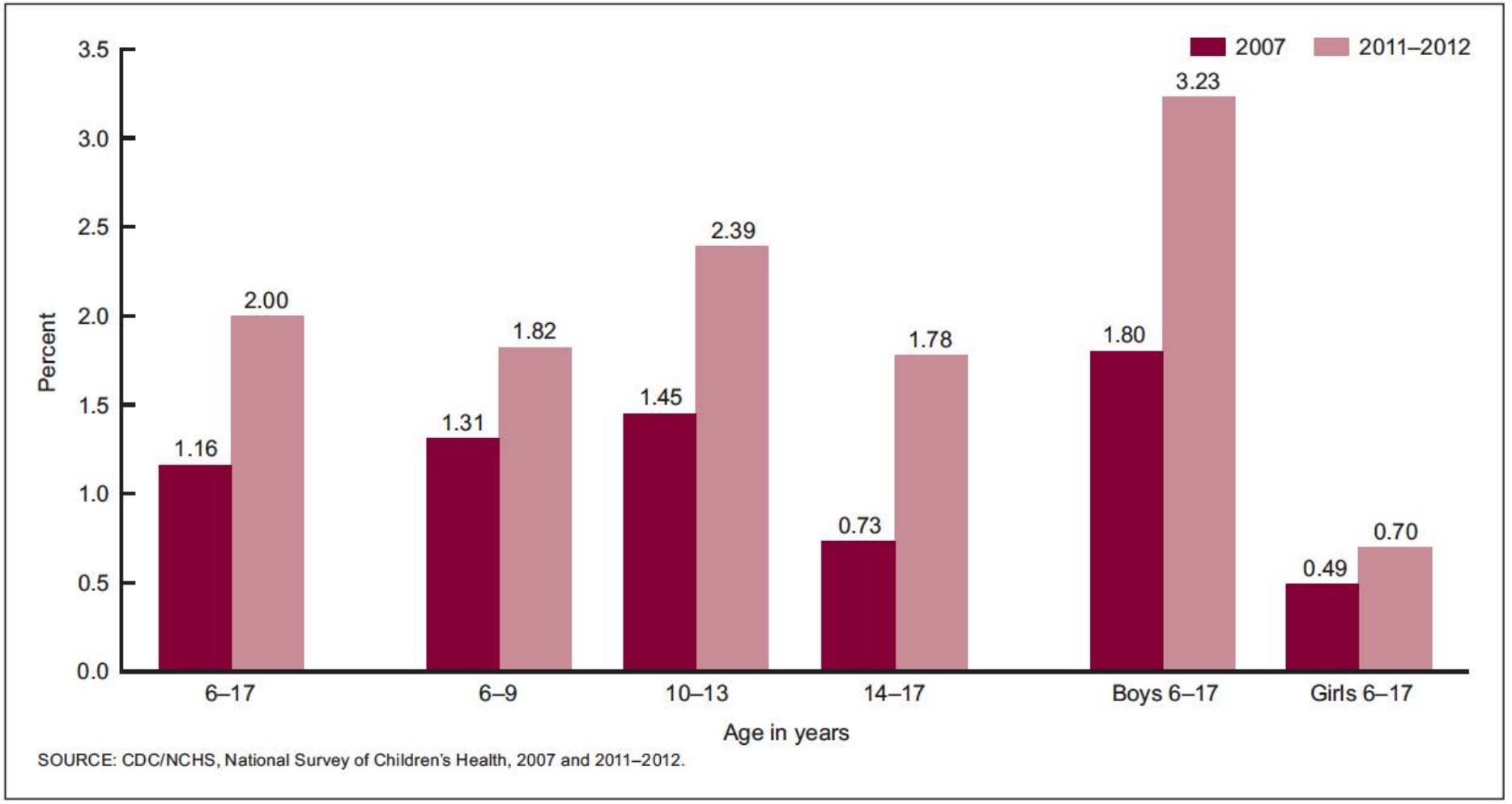


Figure 1. Percentage of children aged 6–17 years with parent-reported autism spectrum disorder, by age group and sex: United States, 2007 and 2011–2012

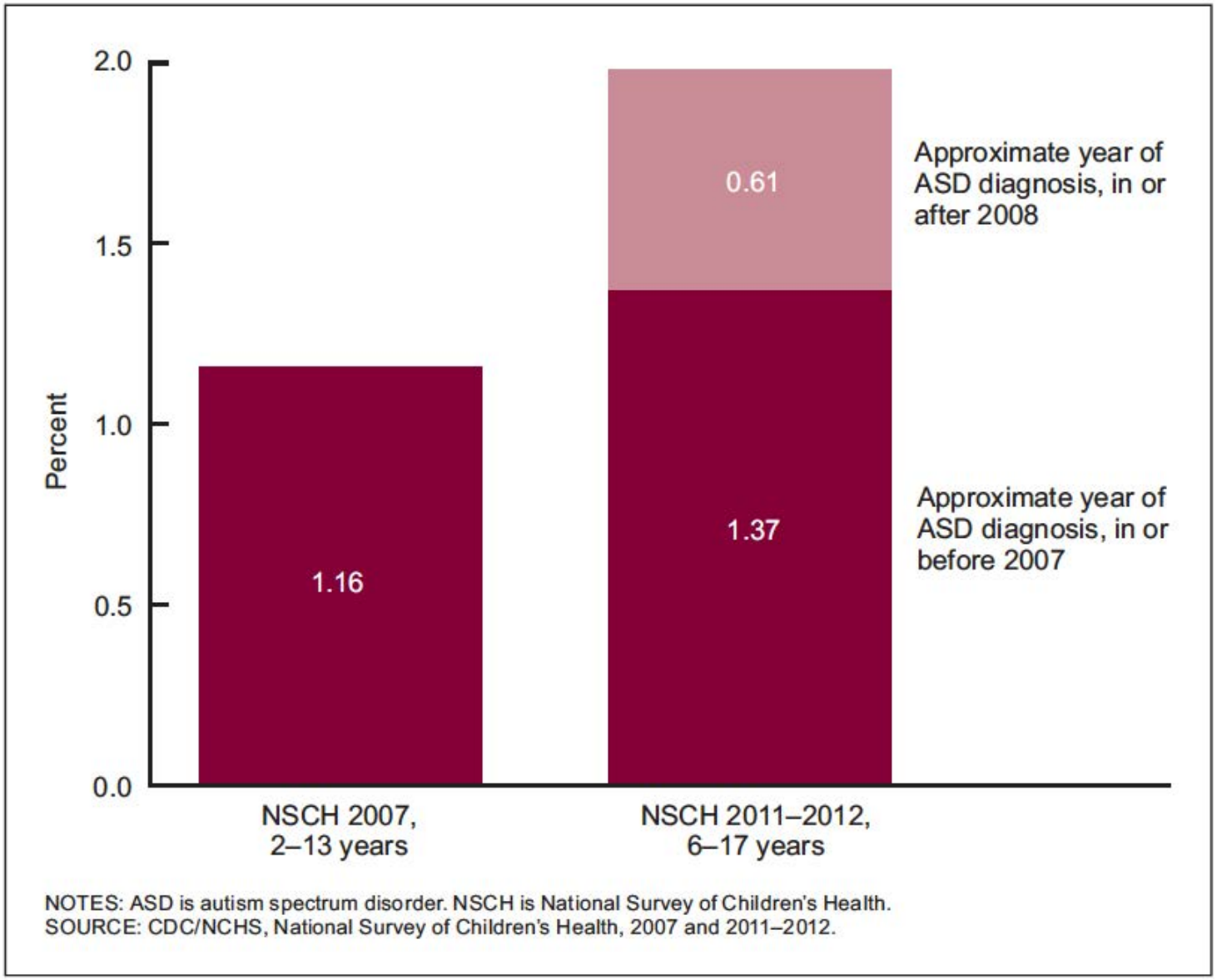
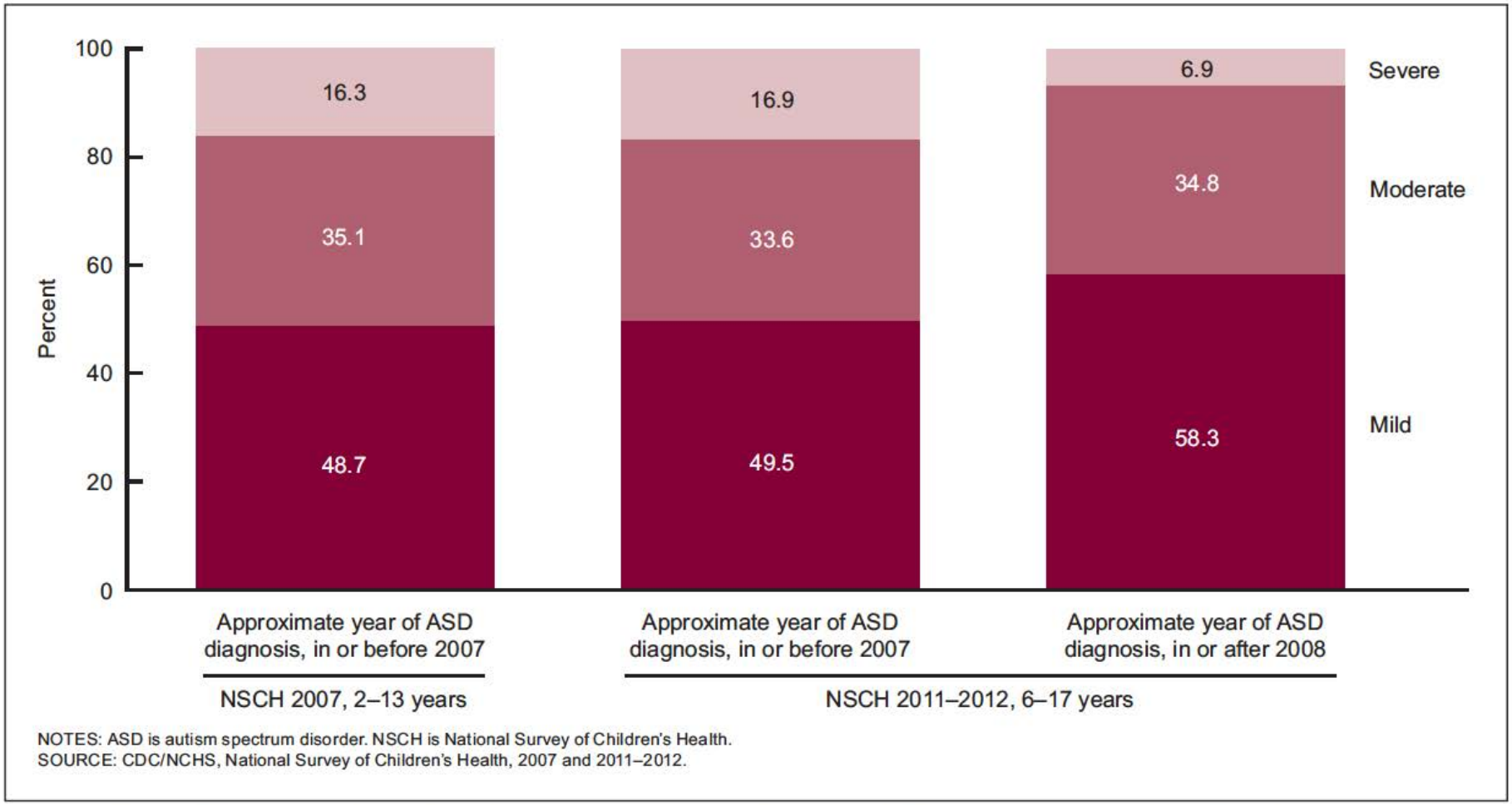


Figure 2. Percentage of children born in 1994–2005 who have parent-reported autism spectrum disorder, by survey and approximate year when child was first diagnosed with ASD: United States, 2007 and 2011–2012



NOTES: ASD is autism spectrum disorder. NSCH is National Survey of Children's Health.
SOURCE: CDC/NCHS, National Survey of Children's Health, 2007 and 2011–2012.

Figure 3. Percent distribution of parent-rated severity of autism spectrum disorder for children born in 1994–2005 who currently have autism spectrum disorder, by survey and approximate year when child was first diagnosed with ASD: United States, 2007 and 2011–2012

Economic cost of autism in the UK

autism © 2009
SAGE Publications
and The National
Autistic Society
Vol 13(3) 317–336; 104246
1362-3613(200905)13:3

MARTIN KNAPP Kings College, London, Institute of Psychiatry, UK and London School of Economics and Political Sciences, UK

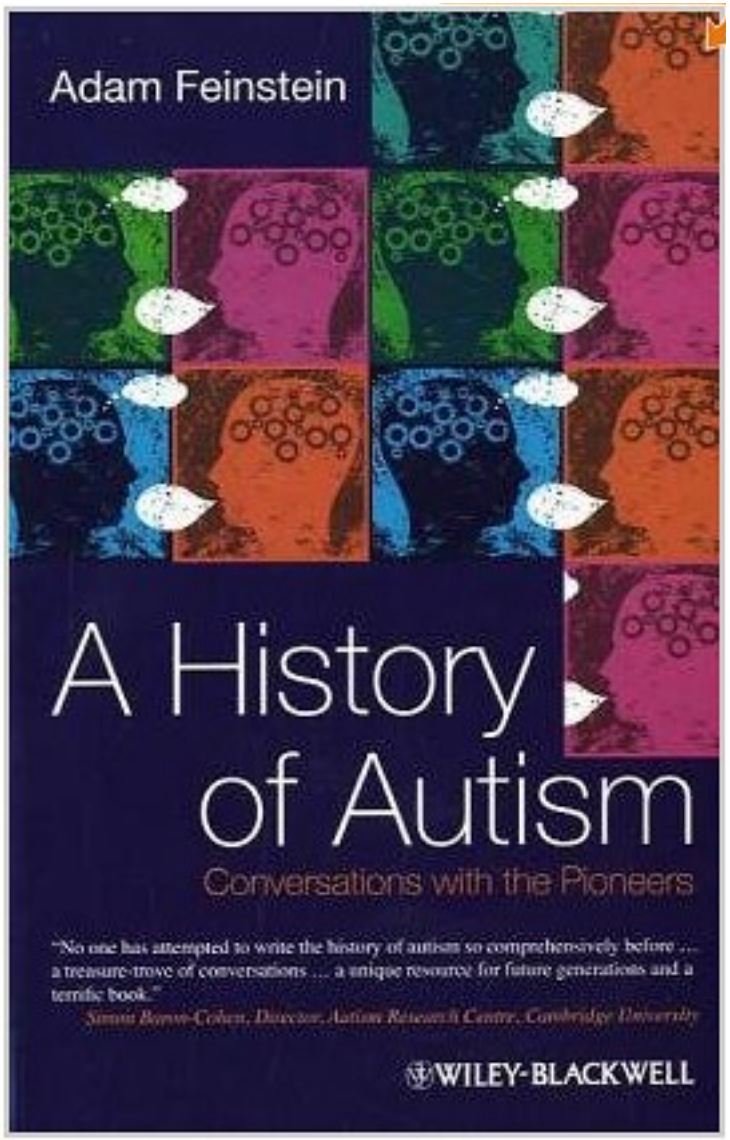
RENÉE ROMEO Kings College, London, Institute of Psychiatry, UK

JENNIFER BEECHAM London School of Economics and Political Science, UK and University of Kent, UK

Still valid?

The costs of supporting children with ASDs were estimated to be £2.7 billion each year. For adults, these costs amount to £25 billion each year.

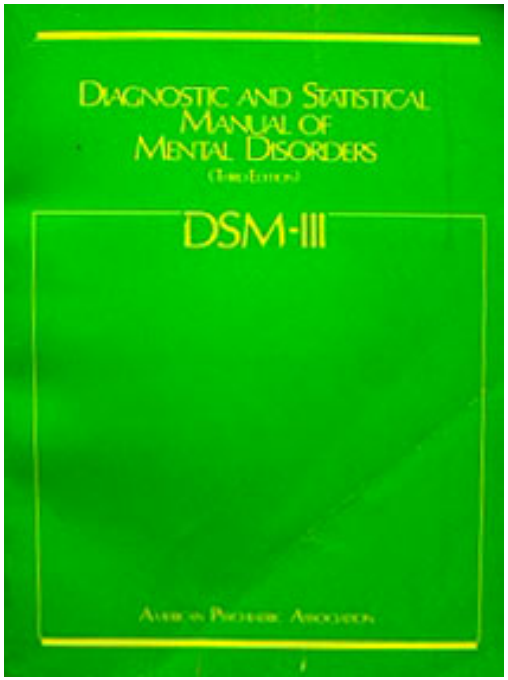
The lifetime cost, after discounting, for someone with ASD and intellectual disability is estimated at approximately **£1.23 million,** and for someone with ASD without intellectual disability is approximately **£0.80 million.**

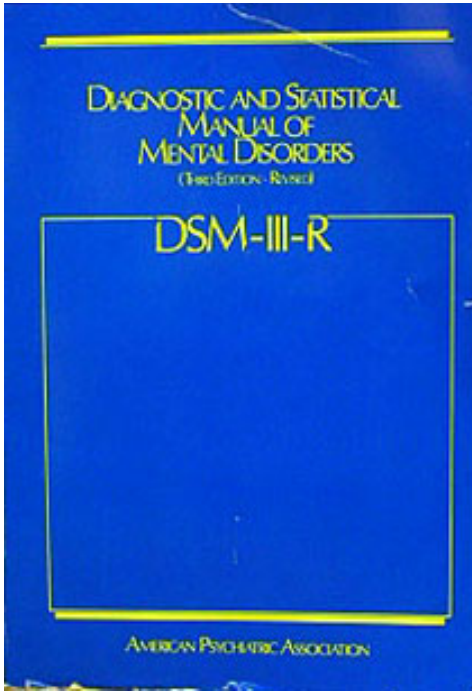

Adam Feinstein
A History of Autism
Conversations with the Pioneers
"No one has attempted to write the history of autism so comprehensively before … a treasure-trove of conversations … a unique resource for future generations and a terrific book."
Simon Baron-Cohen, Director, Autism Research Centre, Cambridge University
WILEY-BLACKWELL

History of diagnostic guidelines

- Autism entered the scientific literature in 1944, but it wasn't until DSM III, published in 1980, that it became a separate diagnostic category.
- DSM-IV, released in 1994 and revised in 2000, laid out six domains that define **autistic disorder** within a triad of impairments
- The DSM-IV introduced **Asperger syndrome,** listing it separately from autism and distinguishing it by the lack of a clinically significant delay in language acquisition and cognitive development.
- **PDD-NOS,** appeared in DSM-IV, usually has limited symptom profile (but in DSM-IV-TR had to have reciprocal social interaction impairment).
- One of the criticisms frequently levelled at DSM-IV is that the expanded diagnostic criteria have led to much higher estimates of prevalence.

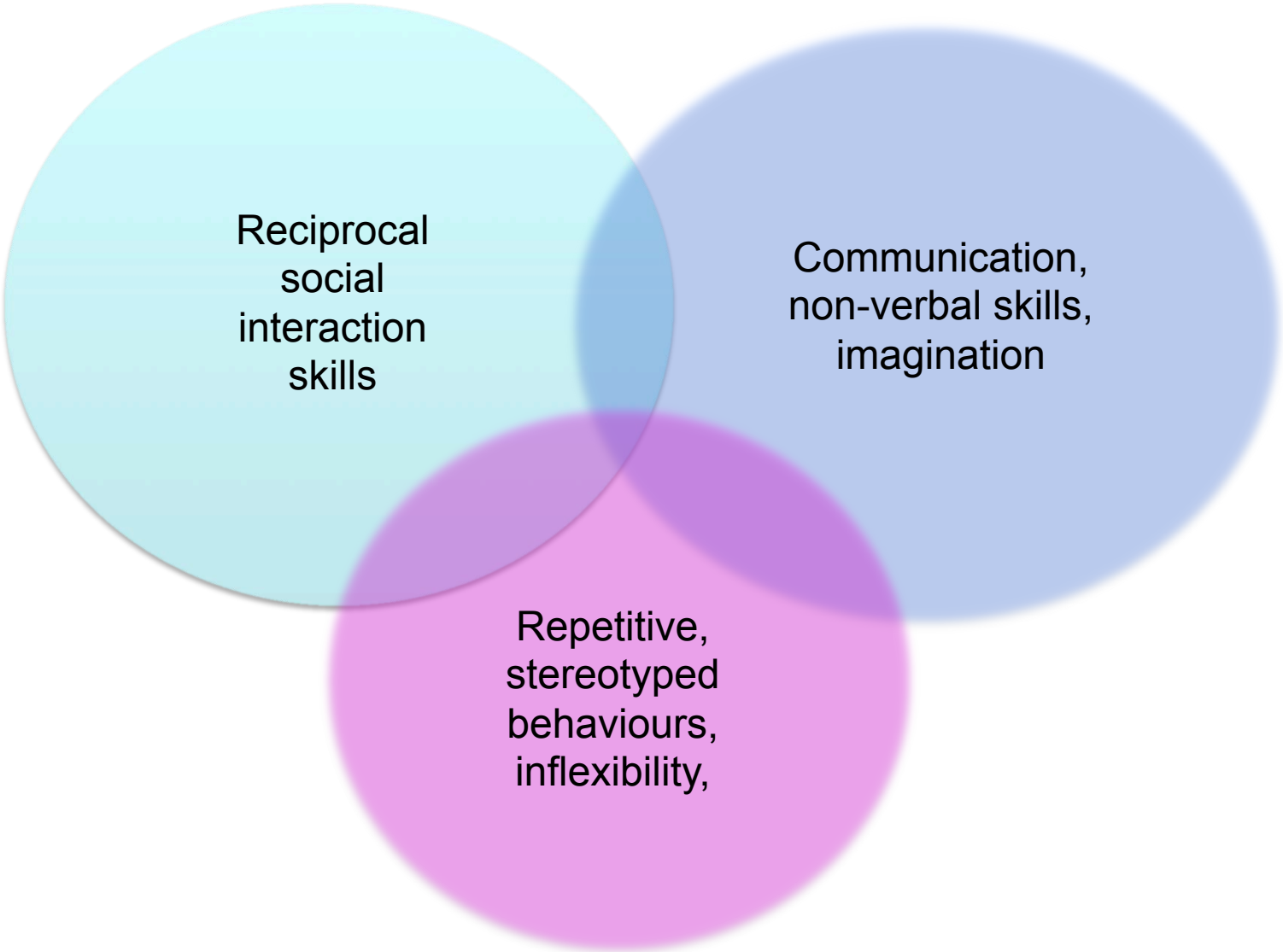
Neither 1980 definition of Autism – DSM-III nor revision in 1987 (DSM-III-R) mentioned Asperger syndrome





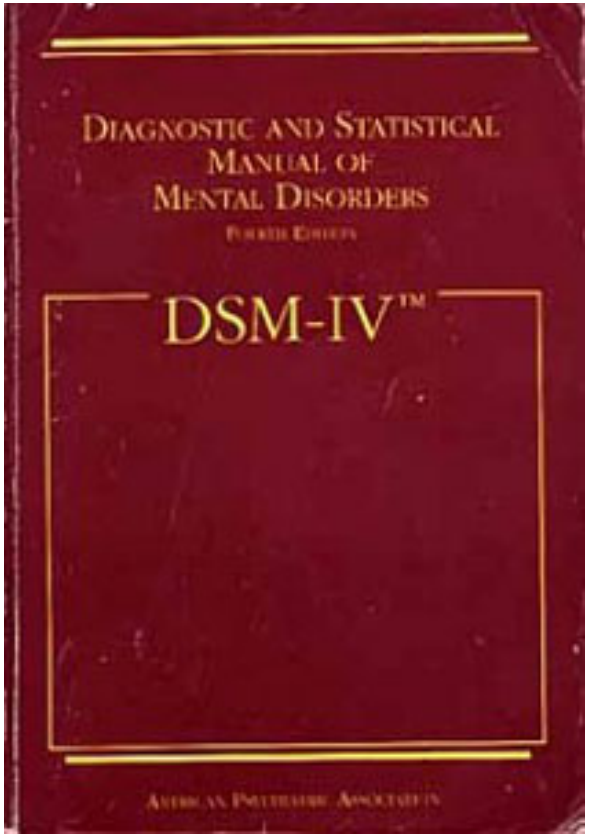
Triad of impairments

ICD-10, DSM-IV-TR



Definition of Asperger's disorder introduced

DSM-IV (1994)



As autism but:

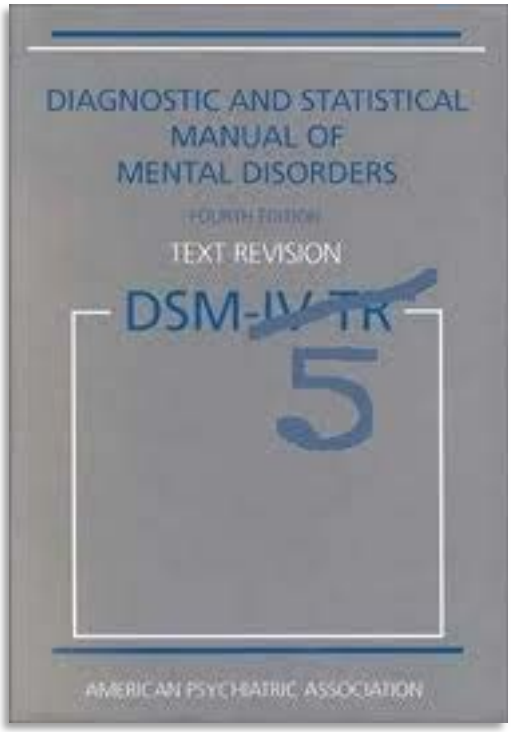
No requirement for a deficit in communication

(IV) No clinically significant general delay in language
(e.g. single words are used by age 2 years, communicative non-echoed phrases are used by age 3 years

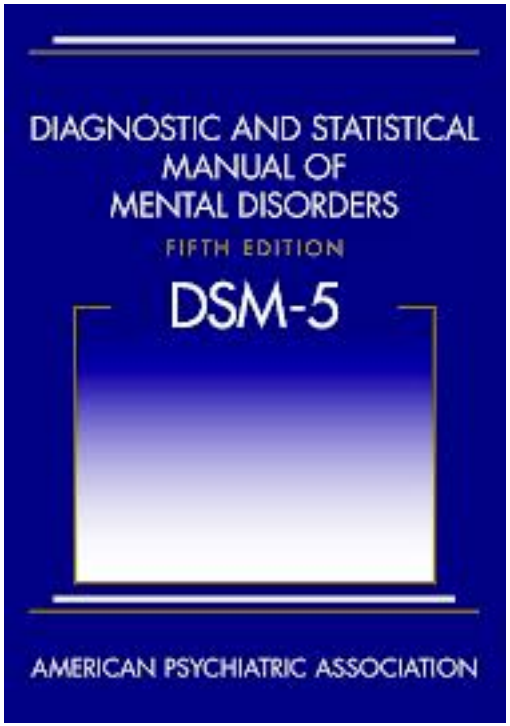
(V) No clinically significant delay:

- in cognitive development
- in the development of age-appropriate self help skills,
- in adaptive behavior (other than in social interaction)
- In curiosity about the environment in childhood.


Autism Spectrum Disorders
Autistic Disorder
Asperger's Disorder
Childhood Disintegrative Disorder
Rett's Disorder
Pervasive Developmental Disorder - Not Otherwise Specified


DIAGNOSTIC AND STATISTICAL
MANUAL OF
MENTAL DISORDERS
FOURTH EDITION
TEXT REVISION
DSM-IV-TR
5
AMERICAN PSYCHIATRIC ASSOCIATION


BACK
TO THE FUTURE


DIAGNOSTIC AND STATISTICAL
MANUAL OF
MENTAL DISORDERS
FIFTH EDITION
DSM-5
AMERICAN PSYCHIATRIC ASSOCIATION

Sensitivity and Specificity of Proposed *DSM-5* Diagnostic Criteria for Autism Spectrum Disorder

James C. McPartland, Ph.D., Brian Reichow, Ph.D., Fred R. Volkmar, M.D.

Reanalysis of a DSM-IV Field Trial:

657 carried a clinical diagnosis of an ASD, and 276 were diagnosed with a non-autistic disorder.

Conclusions:

Revised criteria exclude cognitively able individuals and those with ASDs other than autistic disorder.

JAACAP, April 2012

New Definition of Autism Will Exclude Many, Study Suggests

Todd Heisler/The New York Times

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

CRACKING THE ENIGMA

AN AUTISM RESEARCH BLOG

WE CAN ONLY SEE A SHORT DISTANCE AHEAD BUT WE CAN SEE PLENTY THERE THAT NEEDS TO BE DONE. ALAN TURING.

MONDAY, MARCH 26, 2012

How will DSM 5 affect autism rates?

"If you don't meet the necessary criteria, you're not coming in" Photo by PassportP

In January, at a meeting of the Icelandic Medical Association, Yale researcher, Dr Fred Volkmar gave a presentation of data from a study looking at the implications of changes to autism diagnostic criteria in DSM 5. His conclusion was that many people who are currently diagnosed with autism, Asperger's, or PDD-NOS would not meet the new proposed criteria for autism spectrum disorder in DSM 5.

RECENT POSTS

Autism and intellectual disability in DSM-5

How will DSM 5 affect autism rates?

Conference report: BioAutism 2012

The Adventures of DataThief

Take part in our research on language and auditory processing

Do kids with autism

JON BROCK

I'm a research fellow at the Centre for Cognition and its Disorders at Macquarie University in Sydney. You can see my research profile and publications here.

Take part in our research

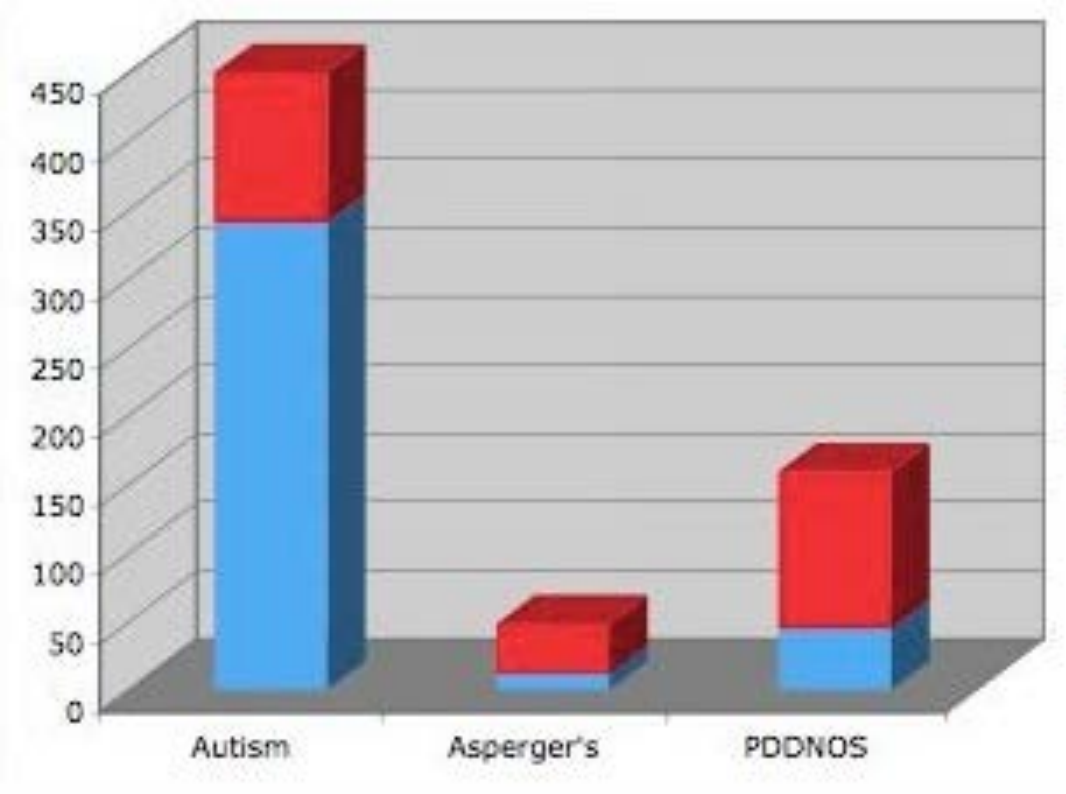
CONNECTIONS

Autism and Empathy - The Caterpillar and The Butterfly: A Love Story

Autism Science Foundation - A summary of the CDC autism prevalence report

Questioning Answers - Six developmental trajectories, autism

Claims for impact of DSM-5 criteria in diagnostic categories defined by DSM-IV

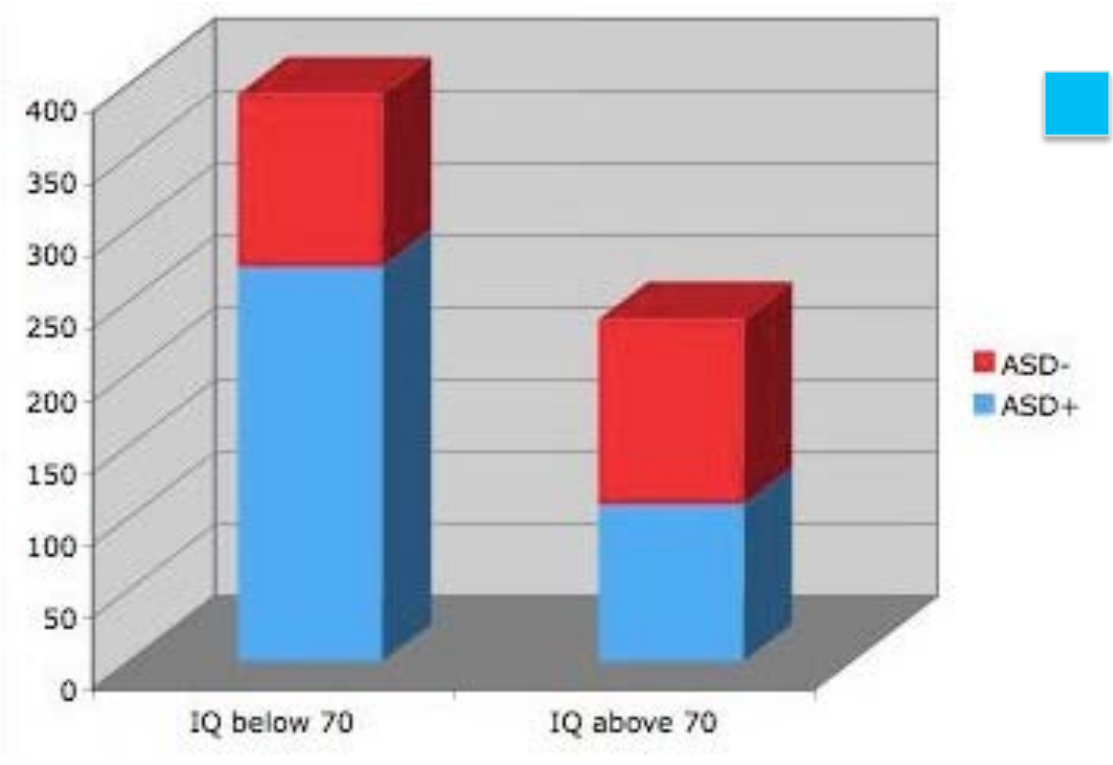


Proportion of DSM-IV sample meeting DSM-5 criteria, by diagnostic category

Figure from Brock blog: McPartland et al, JAACAP, 2012

Sensitivity and Specificity of Proposed *DSM-5* Diagnostic Criteria for Autism Spectrum Disorder

James C. McPartland, Ph.D., Brian Reichow, Ph.D., Fred R. Volkmar, M.D.



Proportion of DSM-IV sample meeting DSM-5 criteria, by diagnostic category

Figures from Brock blog…April 2012

Subtyping autistic disorders

ORIGINAL ARTICLE

ONLINE FIRST

A Multisite Study of the Clinical Diagnosis of Different Autism Spectrum Disorders

Catherine Lord, PhD; Eva Petkova, PhD; Vanessa Hus, MSc; Weijin Gan, MS, MD; Feihan Lu, MA; Donna M. Martin, MD, PhD; Opal Ousley, PhD; Lisa Guy, PhD; Raphael Bernier, PhD; Jennifer Gerdts, MA; Molly Algermissen, PhD; Agnes Whitaker, MD; James S. Sutcliffe, PhD; Zachary Warren, PhD; Ami Klin, PhD; Celine Saulnier, PhD; Ellen Hanson, PhD; Rachel Hundley, PhD; Judith Piggot, MD, PhD; Eric Fombonne, MD; Mandy Steiman, PhD; Judith Miles, MD, PhD; Stephen M. Kanne, PhD; Robin P. Goin-Kochel, PhD; Sarika U. Peters, PhD; Edwin H. Cook, MD; Stephen Guter, MA; Jennifer Tjernagel, MS; Lee Anne Green-Snyder, PhD; Somer Bishop, PhD; Amy Esler, PhD; Katherine Gotham, PhD; Rhiannon Luyster, PhD; Fiona Miller, PhD; Jennifer Olson, PhD; Jennifer Richler, PhD; Susan Risi, PhD

Published March, 2012 in Archives General Psychiatry, this study was very influential in the deliberations of the DSM-5 working party.
It is based on the national survey of participants in the Simons Simplex Collection, with over 2000 children between 4 and 18 years of age

ORIGINAL ARTICLE

ONLINE FIRST

A Multisite Study of the Clinical Diagnosis of Different Autism Spectrum Disorders

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Context: Best-estimate clinical diagnoses of specific autism spectrum disorders (autistic disorder, pervasive developmental disorder–not otherwise specified, and Asperger syndrome) have been used as the diagnostic gold standard, even when information from standardized instruments is available.

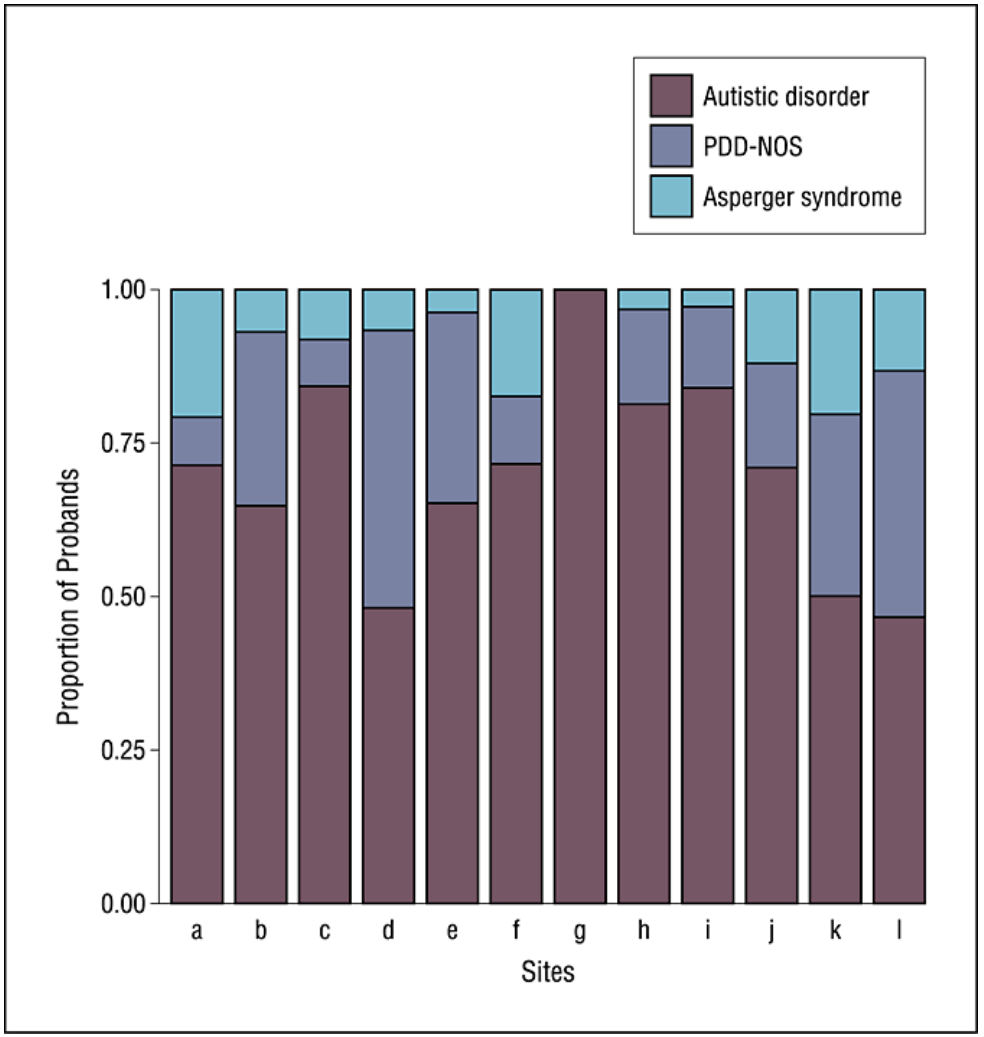
Objective: To determine whether the relationships between behavioral phenotypes and clinical diagnoses of different autism spectrum disorders vary across 12 university-based sites.

Main Outcome Measure: Best-estimate clinical diagnoses predicted by standardized scores from diagnostic, cognitive, and behavioral measures.

Results: Although distributions of scores on standardized measures were similar across sites, significant site differences emerged in best-estimate clinical diagnoses of specific autism spectrum disorders. Relationships between clinical diagnoses and standardized scores, particularly verbal IQ, language level, and core diagnostic features, varied across sites in weighting of information and cutoffs.

Archives General Psychiatry, April 2012

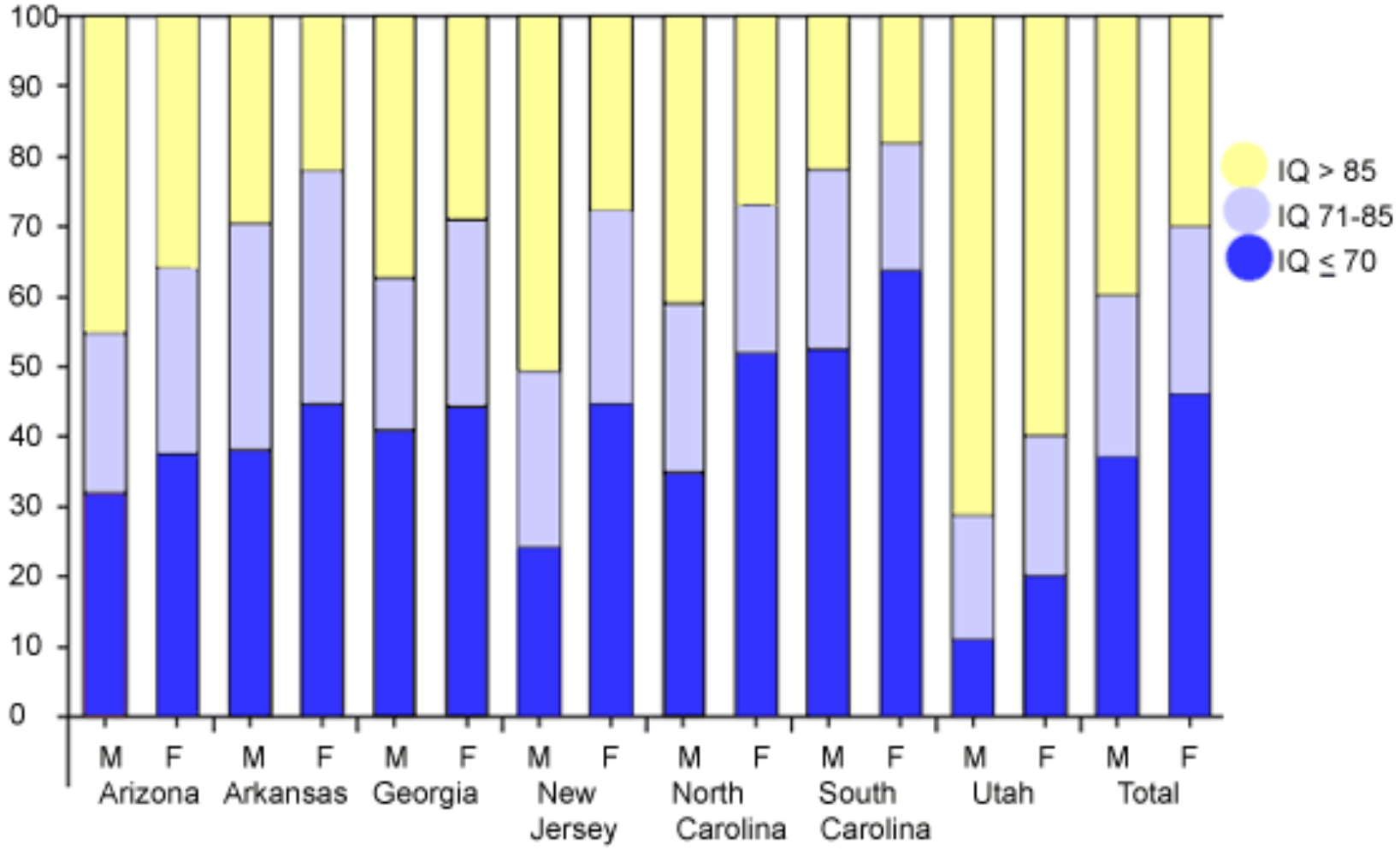
Best-estimate clinical diagnoses across 12 university-based sites (ie, autism service providers) for 2102 probands assigned to 3 autism spectrum disorder diagnostic categories (autistic disorder, Asperger syndrome, and pervasive developmental disorder-not otherwise specified [PDD-NOS])



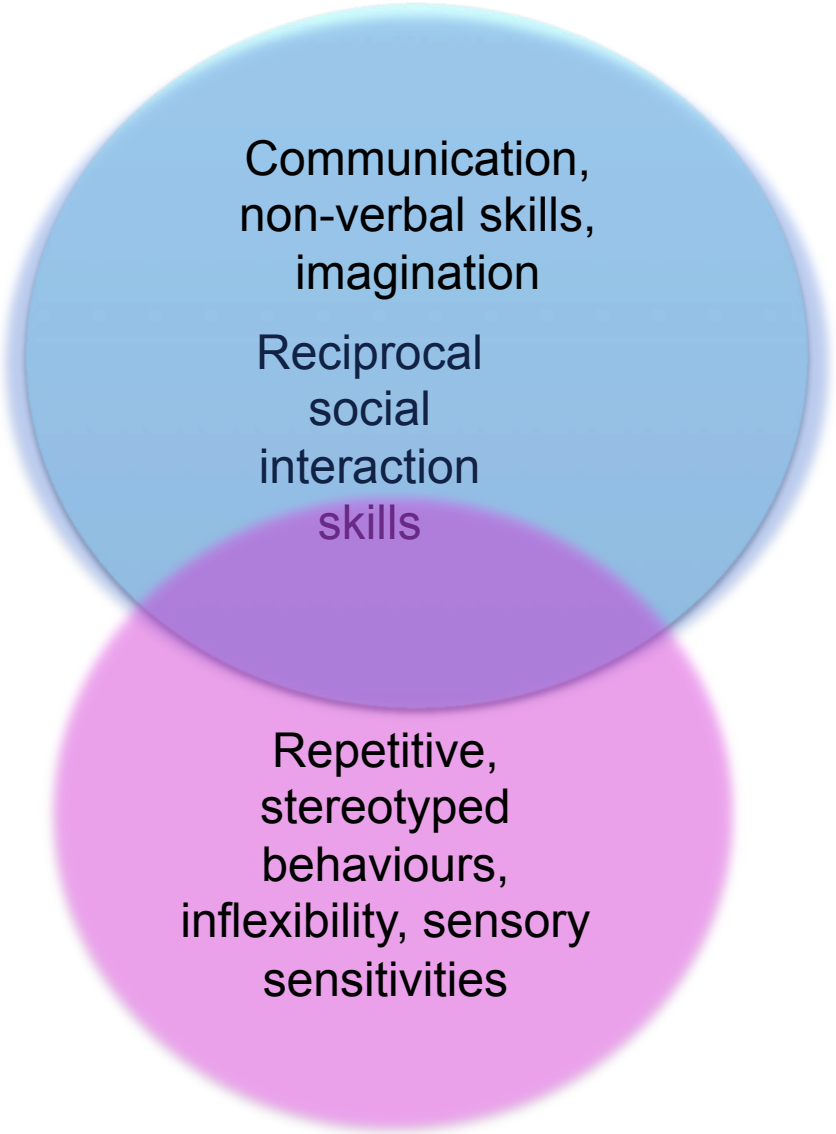
Lord, C. et al. Arch Gen Psychiatry 2012;69:306-313.

ARCHIVES OF GENERAL PSYCHIATRY

Most recent intelligence quotient (IQ) as of age 8 years among children identified with autism spectrum disorders (ASDs) for whom psychometric test data were available,* by site and sex- Autism and Developmental Disabilities Monitoring Network, United States, 2008



DSM-5 –ASD revisions



Autism Spectrum Disorder

Autism Spectrum Disorder
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;
  - ranging from abnormal social approach and failure of normal back and forth conversation through reduced sharing of interests, emotions, and affect and response to total lack of initiation of social interaction,
- 2. Deficits in nonverbal communicative behaviors used for social interaction;
  - ranging from poorly integrated- verbal and nonverbal communication, through abnormalities in eye contact and body-language, or deficits in understanding and use of nonverbal communication, to total lack of facial expression or gestures.
- 3. Deficits in developing, maintaining and understanding relationships,
  - appropriate to developmental level (beyond those with caregivers); ranging from difficulties adjusting behavior to suit different social contexts through difficulties in sharing imaginative play and in making friends to an apparent absence of interest in peers

Specify current severity level (Level 1 to Level 3)

- B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by **at least two** of the following (current behaviour or history):

- 1. Stereotyped or repetitive motor movements, use of objects or **speech;**
  - (such as simple motor stereotypies, echolalia, repetitive use of objects, or idiosyncratic phrases).

- 2. Insistence on sameness, inflexible adherence to routines, ritualized patterns of **verbal** or nonverbal behavior;
  - (including extreme distress at small changes, difficulty with transitions, rigid thinking patterns, greeting rituals, adherence to routes, or very limited range of foods).

- 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 environment;
  - (such as apparent indifference to pain/heat/cold, adverse response to specific sounds or textures, excessive smelling or touching of objects, fascination with lights or movement).

Specify current severity level (Level 1 to Level 3)

2011 draft DSM-5 criteria : Autism Spectrum Disorder **Rationale**

- Deficits in communication and social behaviors **are inseparable** and more accurately considered as a single set of symptoms with contextual and environmental specificities

- Delays in language are not unique nor universal in ASD and are more accurately considered as a factor that influences the clinical symptoms of ASD, rather than defining the ASD diagnosis

- Requiring both criteria to be completely fulfilled (social-communication impairment *and* repetitive and stereotyped behaviors) improves specificity of diagnosis without impairing sensitivity

Scientific American asked astronomer and Hubble Fellow Joshua Peek of Columbia University to code a computer program that would calculate the ways to get a diagnosis under DSM-IV-TR and DSM-5….

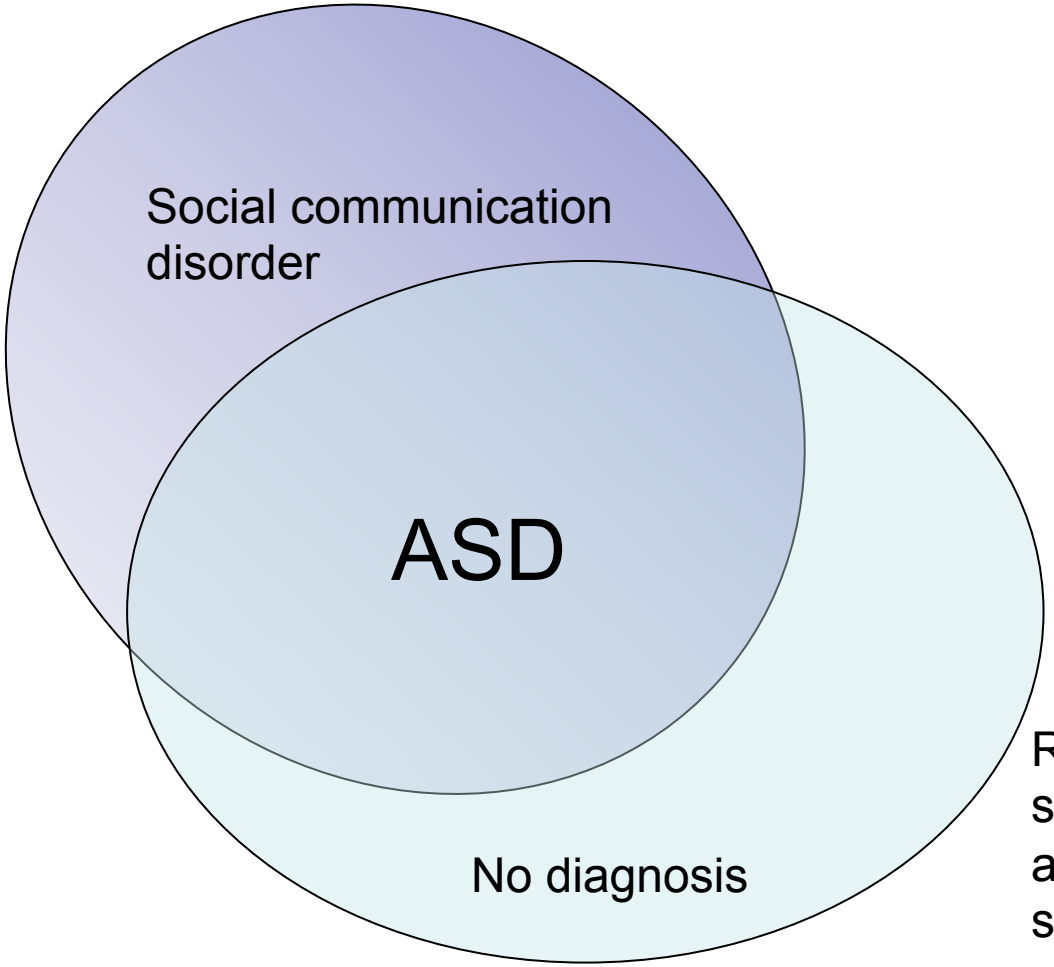
DSM-IV criteria: 12 items in three groups from which you must choose 6, with at least two items from group one and at least one item each from groups two and three.

DSM-5 criteria: 7 items in two groups from which you must choose five, including all three items in group one and at least two of the four items in group two.

Peek's program crunched the numbers: there are 2027 different ways to be diagnosed with autism in DSM-IV and 11 ways to be diagnosed with autism in DSM-5.

Revised conceptualization of the autism spectrum

Social – communication deficits



Repetitive, stereotyped interests and behaviours, sensory sensitivities

Skuse, 2012, JAACAP

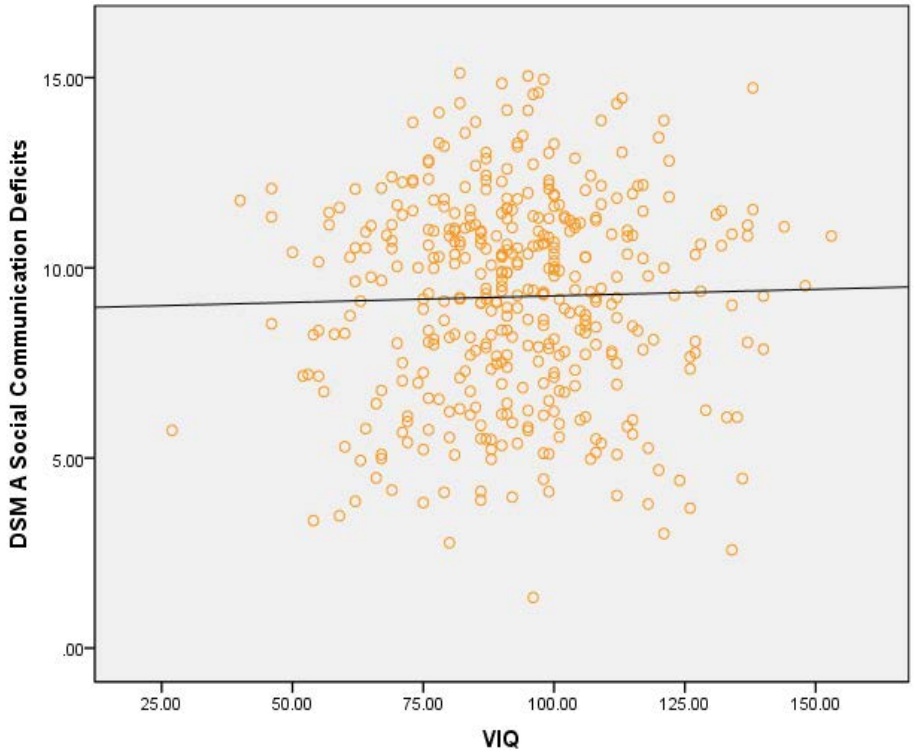
Agreement by DSM-IV & DSM-5 diagnosis

	Broad phenotype	Autism	Asperger syndrome	PDD-NOS
	N (column %)	N (column %)	N (column %)	N (column %)
DSM-5 ASD-	156 (68%)	17 (4%)	5 (3%)	38 (23%)
DSM-5 ASD+	75 (32%)	179 (96%)	138 (97%)	129 (77%)
Sensitivity	**-**	**.96**	**.97**	**.77**
Specificity	**-**	**.68**	**.68**	**.68**

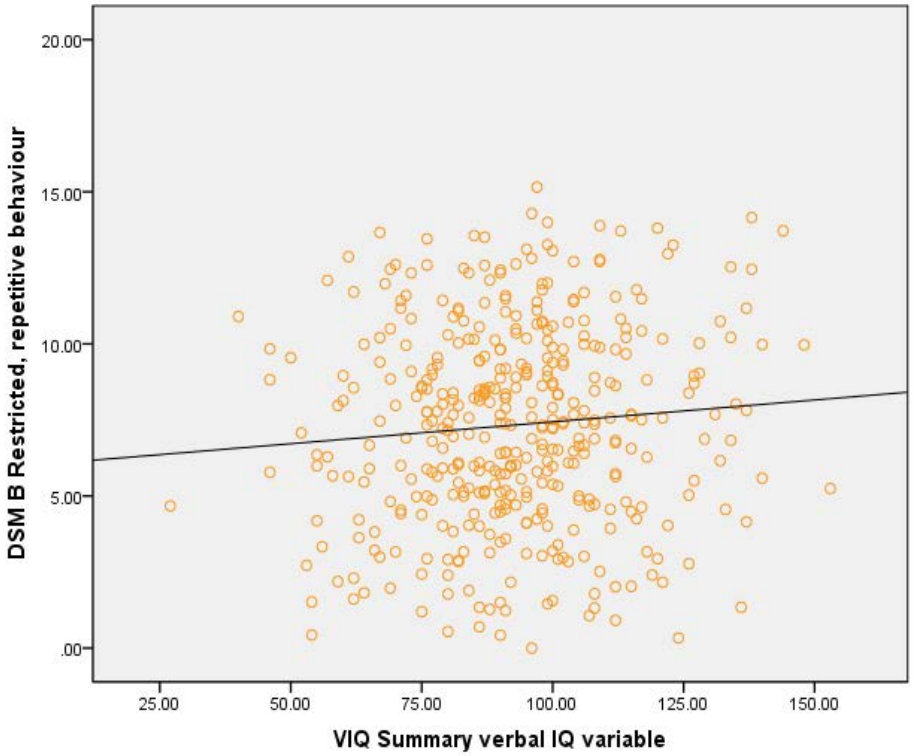
- Sensitivity and specificity are much poorer for PDD-NOS
- ~25% of children with a current PDD-NOS diagnosis could lose that diagnosis under DSM-5 criteria

No relationship between DSM-5 symptom domains and IQ

Social Communication



Repetitive, restricted behaviour



EDITORIAL

DSM-5's Conceptualization of Autistic Disorders

David H. Skuse, M.D.

- What does DSM-5 get right?
  - Takes a dimensional approach to diagnosis of ASD
  - Combines social reciprocity and communication subscales
  - Introduces sensory sensitivities as criteria *(both positive and negative)*
  - Removes 'impaired imagination' from list of core symptoms *(note, the key issue is whether this can be used socially)*
  - Reassigns stereotyped/repetitive language as a form of RRB *(note this is a complex issue, some such language is social)*

EDITORIAL

DSM-5's Conceptualization of Autistic Disorders

David H. Skuse, M.D.

- What else does DSM-5 get right?
  - Removes criterion of early language delay
  - Drops the diagnosis of Asperger syndrome
  - Removes exclusion criteria for comorbidity such as ADHD

- What does DSM-5 get wrong?
  - Introduces a new category of Social Communication Disorder as a residual diagnosis for children without sufficient RRSB!

Social Communication Disorder

- Deficits in communication for **social purposes**, including greeting and sharing information
- Communication style not matched to **social context**
- Difficulty **following rules** for conversation or story-telling, taking turns in conversation, rephrasing to improve clarity of communication
- Poor **regulation of interaction** through verbal and non-verbal cues
- Difficulties with **implicit understanding** of verbal and non-verbal cues (e.g. idioms, hints, humor)


ASD
Miller
lite

RESEARCH ARTICLE

Toward Specifying Pervasive Developmental Disorder—Not Otherwise Specified

William Mandy, Tony Charman, Jane Gilmour, and David Skuse

Pervasive developmental disorder—not otherwise specified (PDD-NOS) is the most common and least satisfactory of the PDD diagnoses. It is not formally operationalized, which limits its reliability and has hampered attempts to assess its validity. We aimed, first, to improve the reliability and replicability of PDD-NOS by operationalizing its DSM-IV-TR description and, second, to test its validity through comparison with autistic disorder (AD) and Asperger's disorder (AsD). In a sample of 256 young people (mean age = 9.1 years) we used Developmental, Diagnostic and Dimensional (3Di) algorithmic analysis to classify DSM-IV-TR AD ($n = 97$), AsD ($n = 93$) and PDD-NOS ($n = 66$). Groups were compared on independent measures of core PDD symptomatology, associated autistic features, and intelligence. Contrary to the assumption that PDD-NOS is heterogeneous, almost all (97%) of those with PDD-NOS had one distinct symptom pattern, namely impairments in social reciprocity and communication, without significant repetitive and stereotyped behaviors (RSB). Compared to AD and AsD, they had comparably severe but more circumscribed social communication difficulties, with fewer non-social features of autism, such as sensory, feeding and visuo-spatial problems. These individuals appear to have a distinct variant of autism that does not merely sit at the less severe end of the same continuum of symptoms. The current draft guidelines for DSM-V, which mandate the presence of RSBs for any PDD diagnosis, would exclude such people from the autistic spectrum.

Keywords: pervasive developmental disorder—not otherwise specified (PDD-NOS); Autistic disorder; Asperger's disorder; autism spectrum disorder; diagnostic and statistical manual (DSM)

97% of all PDD-NOS in clinical sample of ASD lacked repetitive/stereotyped behaviours and so would be eligible for diagnosis of SCD

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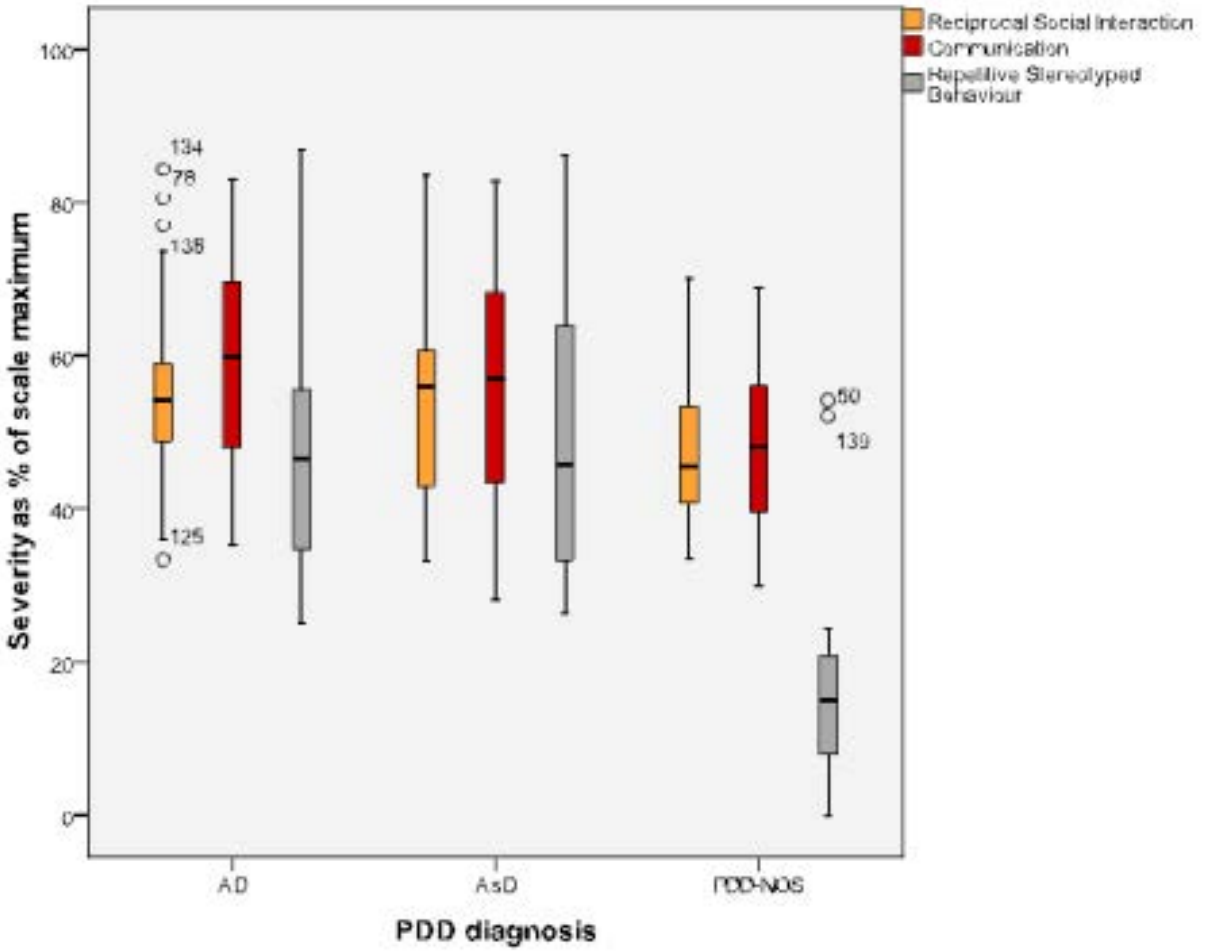
In PDD-NOS severity of RSB is reduced



Autism Res. 2011

DSM-5's Conceptualization of Autistic Disorders

David H. Skuse, M.D.

Asked, can criteria for SCD be defined in such a way that they do not amount to ASD equivalent "social/communication deficits" in the absence of (an arbitrary number of) fixated interests and repetitive behaviors?

Answer: *no – the published criteria simply state there should be no history of 'any' restrictive, repetitive patterns of behaviour, interests or activities*

Asked, where is the evidence that the etiology, appropriate management, or prognosis of the new condition is different from that of ASD?

Answer: *not yet.*