# The context of DSM5 : Where did it come from and where may it lead us?

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# Conflict of Interest

- I receive royalties from diagnostic instruments from Western Psychological Services including the ADI-R, ADOS2 and SCQ

- I have research funding from NIMH, NICHD, HRSA, Autism Speaks and the Simons Foundation.

# Outline of talk

- General issues in diagnosis of Autism Spectrum Disorder (ASD)
- Changes in DSM 5 ASD criteria
- New Pragmatic Social Communication Disorder
- Specifiers
- Assessment of severity through need for support

- General comments; implications for services

- Not discussing intellectual disabilities, communication disorders or learning disabilities but am happy to answer questions about them later

# Clinical challenges of Autism Spectrum Disorders (ASD)

- Individual differences
  - In severity of ASD symptoms
  - In families' priorities, resources, needs
  - In other aspects of development
- Importance of context in ASD symptoms
- Lack of biological markers
- Availability of medical treatments that can have real, but relatively minor effects on co-occurring conditions
- Lack of adequate funding for services and knowledge of what works for whom

# Other burning issues in ASD

- How broad should diagnoses be?
- Can symptom counts work as well as more integrated systems in providing meaningful behavioral diagnoses or descriptions?
- Where does impairment fit in diagnostic criteria?
- What is the balance between requiring a carefully made but expensive diagnosis resulting in good sensitivity and specificity and having something cheap and quick with very poor specificity?
- What is the value of a diagnostic assessment? How does a diagnostic assessment contribute to caregiver and self understanding and treatment planning?

Autism is a developmental disorder:
What is manifested as autism changes with development
Development is affected by having autism; ASD as a disorder of learning

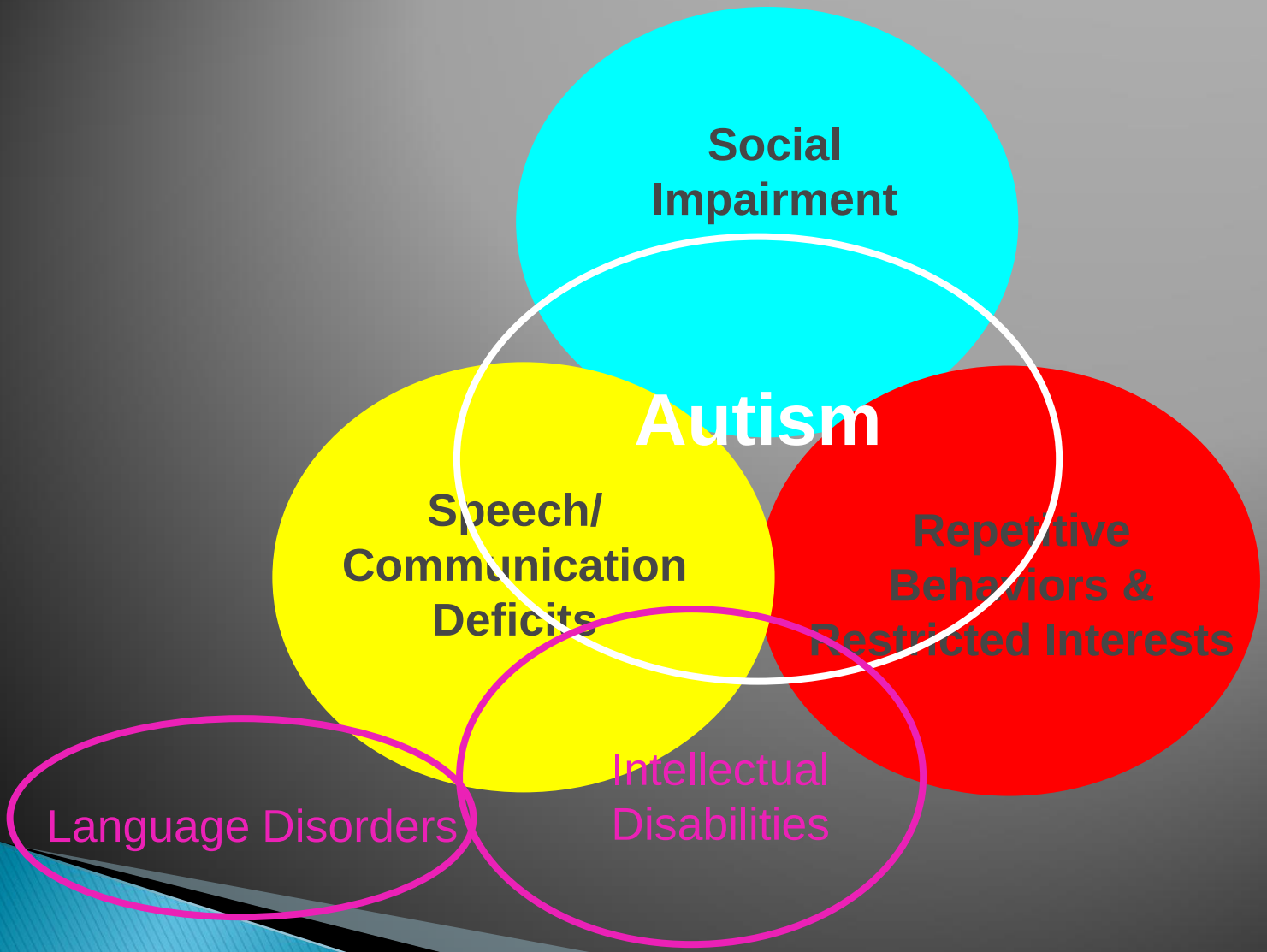
- Both positive (abnormal) behaviors, and negative (the absence of normal) behaviors are required to make a diagnosis of ASD. In research, these “items” often group together.

- This means that developmental level (the age level at which a person is functioning) and situational effects (in what kind of circumstances does the child or adult behave like this?) both have significant effects on diagnostic judgments.

# Where were we in diagnosis before DSM5?

- 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 autism were generally stable across many years.
- However, diagnoses of specific PDDs were problematic.

# Pervasive Developmental Disorders



# DSM 5 Commmittee on Neurodevelopmental Disorders

- Susan Swedo, M.D. , pediatrician and chair
- Gillian Baird, M.D., developmental pediatrician
- Edwin Cook Jr, M.D., child psychiatrist
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- Rosemary Tannock, Ph.,D., pediatric neuropsychologist
- Amy Wetherby, Ph.D., speech-language pathologist
- Harry Wright, M.D., child psychiatrist

# DSM5 process

- Committee and chair appointed by American Psychiatric Association
- Weekly conference calls; face to face meetings about every 3 months for 4 years
- Decisions made through discussion, drafting of proposals, consensus in most cases
- Various advisors consulted, including autism self-advocates and other experts
- Drafts posted and comments reviewed
- Drafts written by individuals, reviewed by subcommittees and then full committee and DSM5 review groups

# Research aspects of DSM5

- Among our committee, we had access to several large datasets predominantly of clinical referrals for ASD and research participants in ASD projects, but also some individuals with related, but non-ASD diagnoses (existing data)
- Iterative analyses were run, reviewed and re-run to test alternative aspects of drafts, but in the end committee discussion preempted data
- DSM5 field trials were conducted with draft criteria; committee was given results

# Decision process

- Sensitivity always trumps specificity in every disorder
- No axes (as determined by APA)
- All disorders must have severity indices (as per APA)
- No specification of research standards or methods
- General interest in dimensions but no agreement about what they should be or how to measure them
- For ICD 11, commitment to primary care

# Goals in revising DSM5 criteria

Do not to change who is included

Make the framework more useful for all ages, all developmental levels and all degrees of severity where there is impairment

Make sure that the criteria do describe ASD and don't describe many people who don't have ASD

Allow separate ways of describing behaviors and noting etiology and associated conditions

# Changes in DSM 5

- 1. One spectrum of autistic disorders called Autism Spectrum Disorder (ASD) defined purely by behaviors
  - No categorical differentiation among autism, PDD-NOS, Asperger Syndrome, Childhood Disintegrative Disorder
  - No categorical differentiation within ASD among disorders by etiology (Rett Syndrome, Fragile X, other known genetic disorders)
  - These differences are now noted through specifiers and modifiers

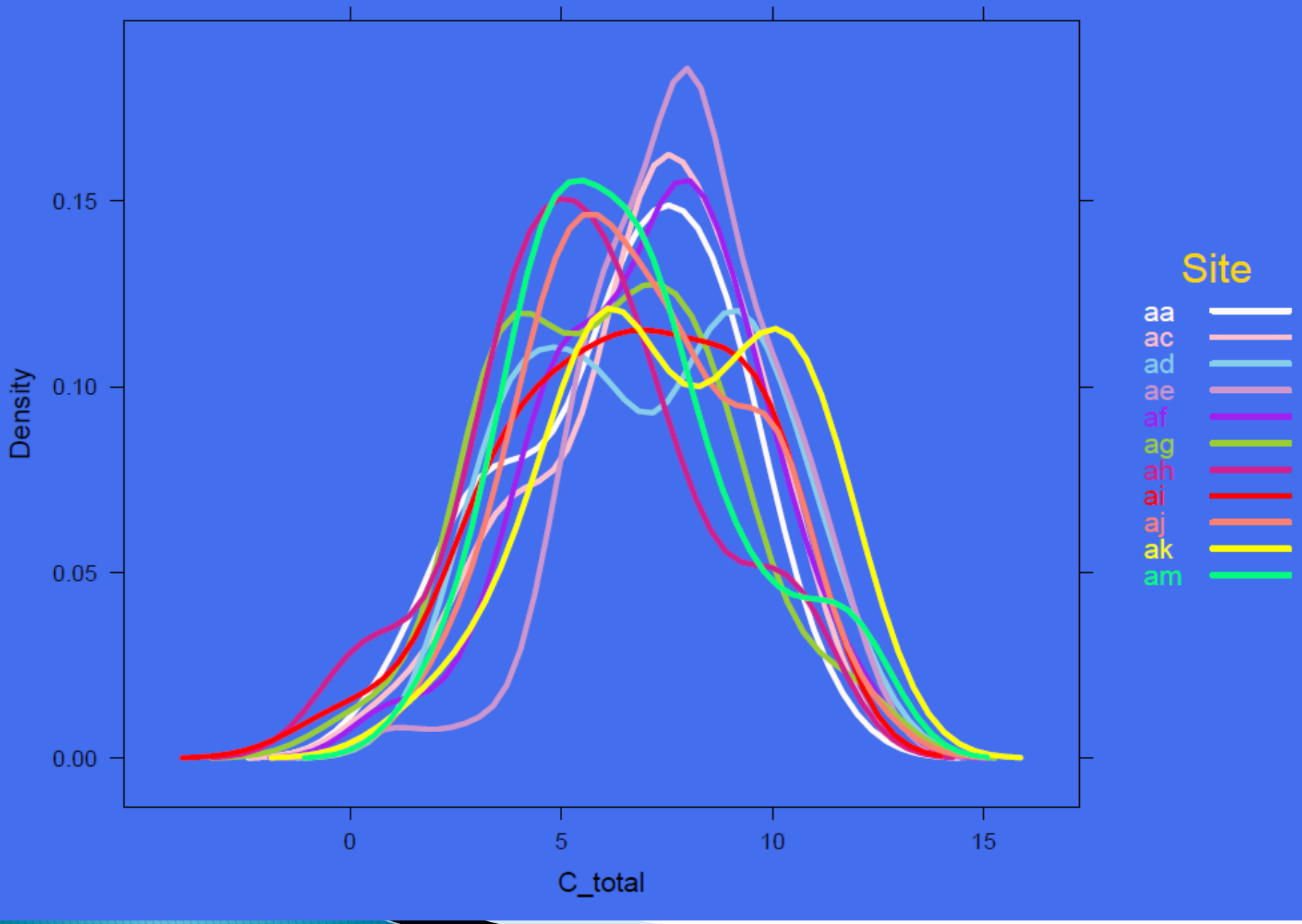
# Many reasons to include Asperger Syndrome and PDD-NOS within one ASD diagnosis

- 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

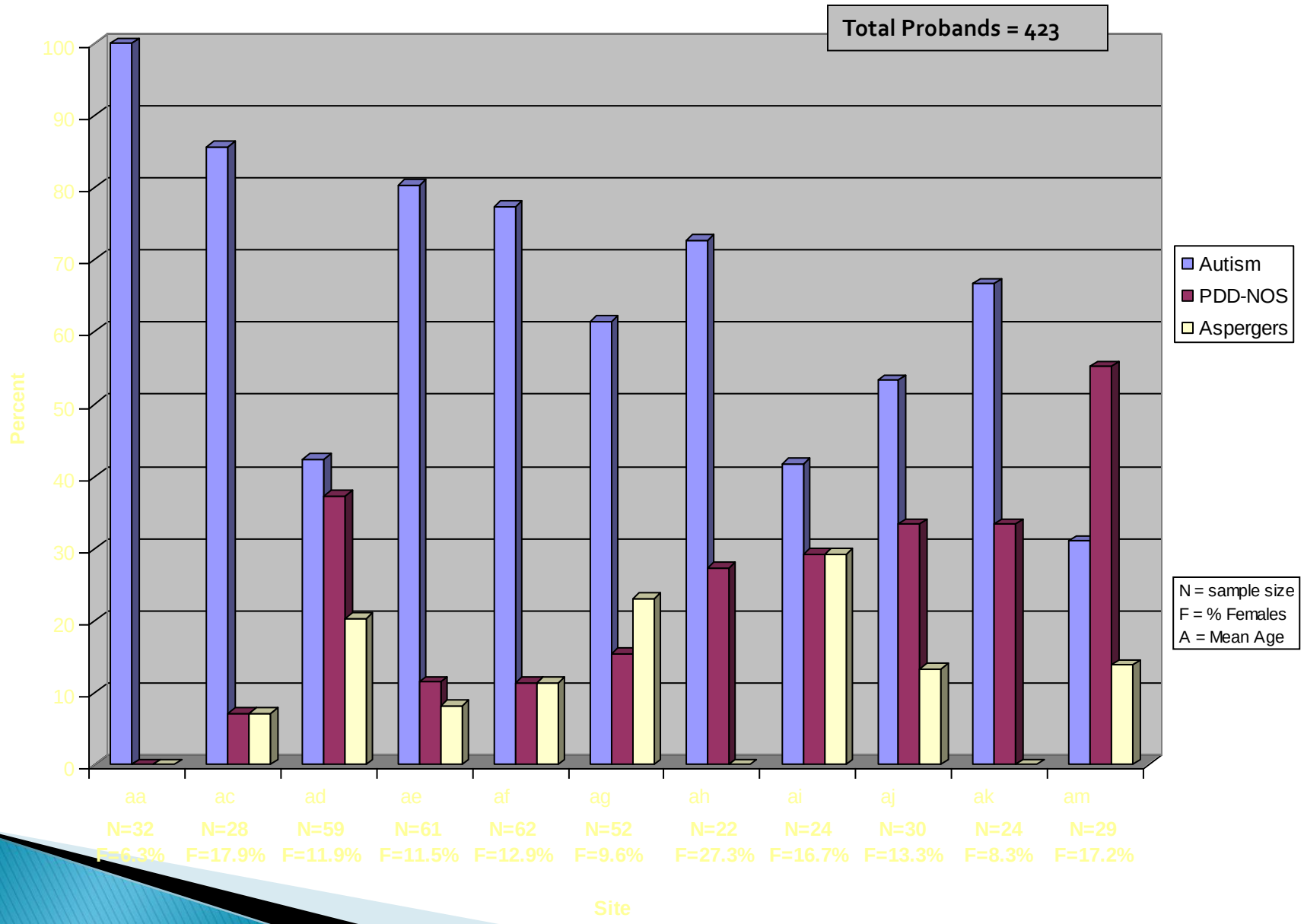
# Simons Simplex Collection

- Over 2700 validated singletons with ASD;
- 9000 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
- Fischbach & Lord, 2011, Neuron; Lord et al., 2012, Arch Gen Psychiatry

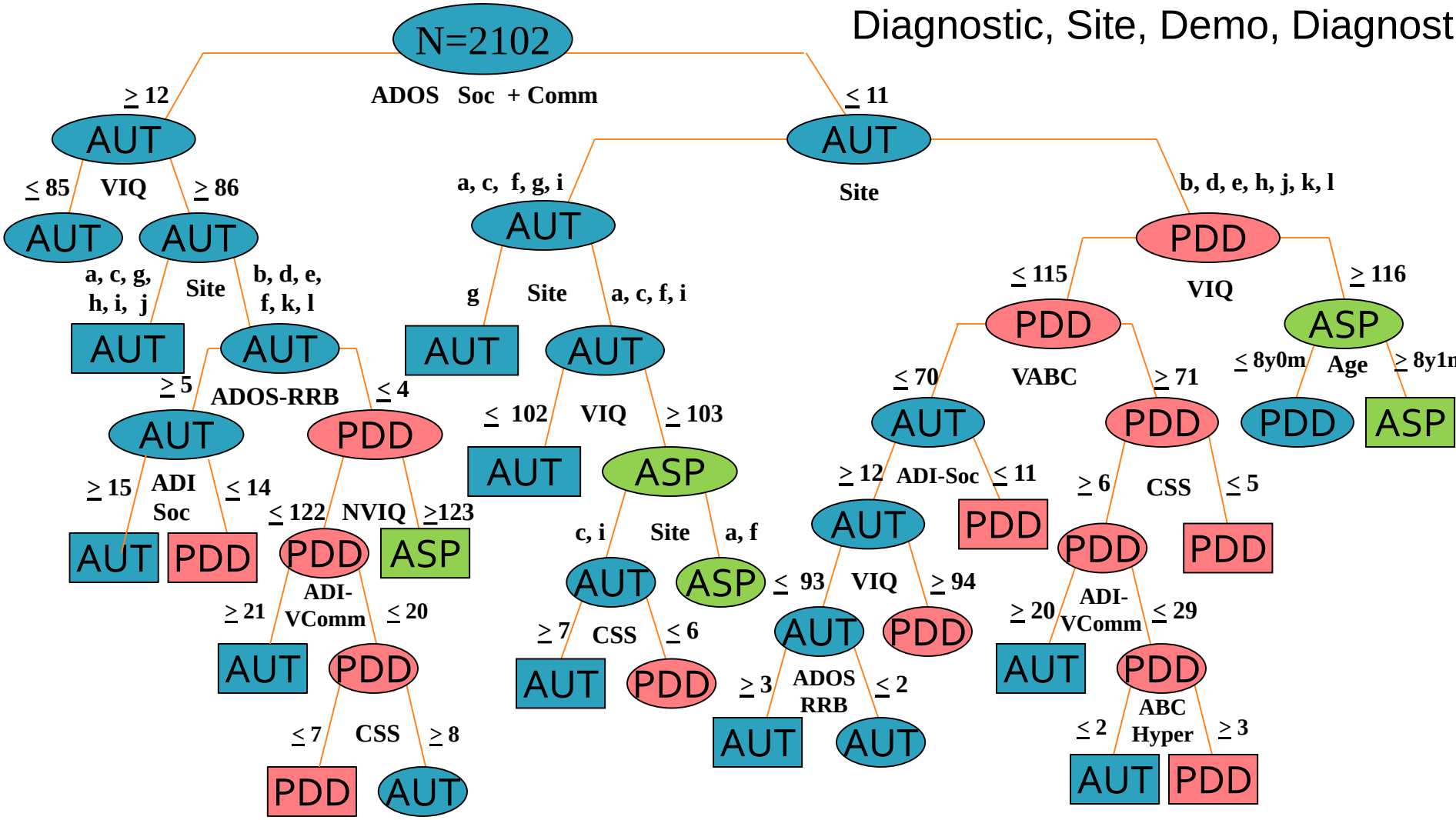
# ADI-R RRB Domain Scores



# ASD Distribution of Probands



# Diagnostic, Site, Demo, Diagnosti




*The Simons Simplex Collection*

Lord et al. (in press)

# Predictors of various ASD diagnoses by site

| | a | b | c | d | e | f | h | i | l | k |
|---|---|---|---|---|---|---|---|---|---|---|
| 1st split | VIQ | ADOS SocAff | ADOS Soc+Com | VIQ | ADOS Soc+Com | VIQ | ADOS RRB | ADOS SocAff | ADOS SocAff | VIQ |
| | VIQ | VIQ | ADOS Soc+Com | ADOS RRB | ADOS Soc+Com | VIQ | ADOS RRB | VIQ | ADOS Soc+Com | ADOS Soc+Com |
| | Vineland | ADI NV-Comm | CSS | ADOS Soc+Com | CSS | ADOS Soc+Com | ADOS Soc+Com | ADOS Soc+Com | CSS | Vineland |
| | ADOS Soc+Com | ADOS RRB | VIQ | VIQ | NVIQ | NVIQ | Mat Educ | ADOS Mod | VIQ | ADI Social |
| | CSS | ADOS Soc+Com | ADOS RRB | ADOS Mod | VIQ | CSS | CSS | NVIQ | ADOS Mod | VIQ |

| | a | b | c | d | e | f | h | i | l | k |
|---|---|---|---|---|---|---|---|---|---|---|
| 2nd split | ADOS RRB | NVIQ | ADI RRB | CSS | ADOS RRB | ADOS RRB | ADOS Mod | Vineland | ADOS RRB | NVIQ |
| | Vineland | NVIQ | CSS | ADOS Soc+Com | CSS | NVIQ | ADOS Soc+Com | NVIQ | CSS | CSS |
| | NVIQ | ADOS Mod | ADOS RRB | ADOS Mod | ADOS RRB | ADOS Soc+Com | VIQ | ADOS Mod | VIQ | ADOS RRB |
| | ADOS Mod | Vineland | VIQ | VIQ | VIQ | ADOS RRB | CSS | ADOS Soc+Com | ADI Social | VIQ |
| | ADI Social | ADOS Soc+Com | ADI NV-Comm | NVIQ | ADI Social | CSS | ADI RRB | ADOS Soc+Com | ADOS RRB | ADOS Mod |

# Care needs to be taken

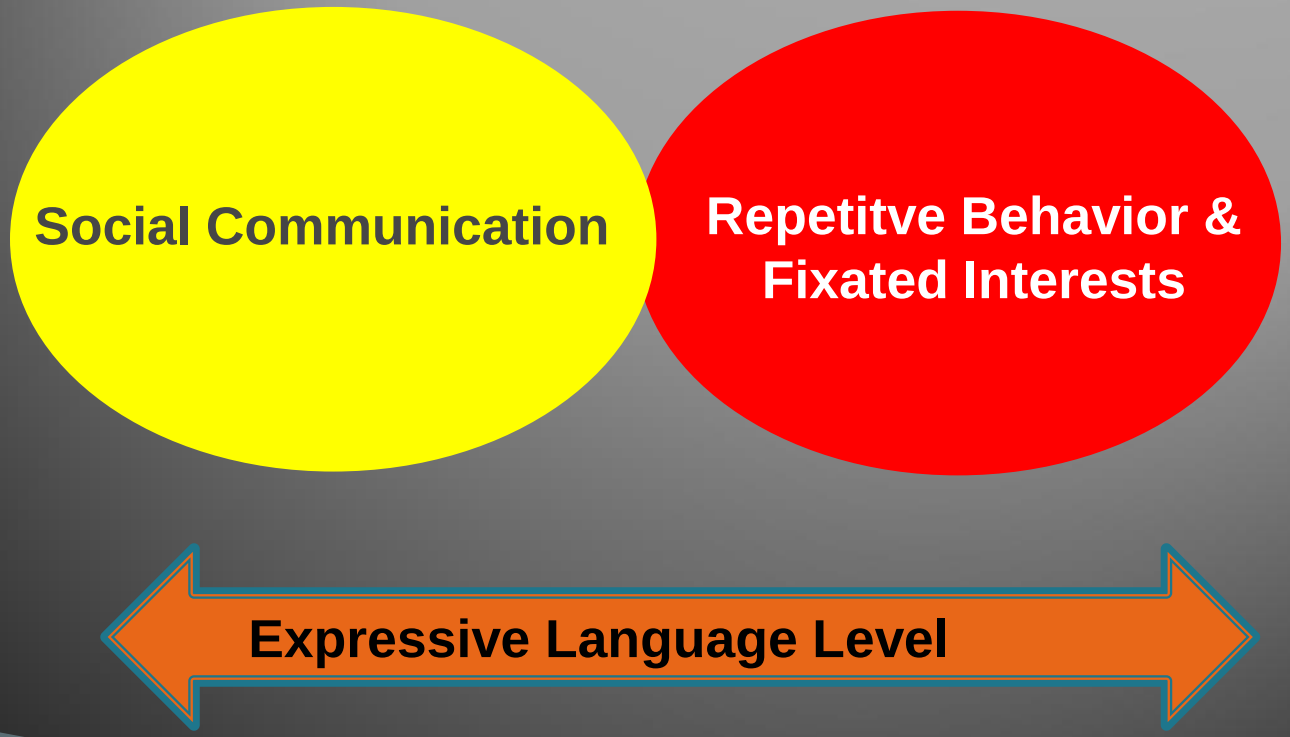
- That people with diagnoses of Asperger Syndrome or PDD-NOS do not lose services because of being included in ASD
- That people who prefer the term Asperger Syndrome to refer to themselves can use it
- That the ranges of skill levels and abilities within ASD are not underestimated

# 2. How many domains?

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

- Social communication
- Fixated interests and repetitive behaviors (RRBs)

# New Domains in DSM5



# 3. For social-communication, criteria must be met within EACH subdomain currently or by history

- ***Deficits in social-emotional reciprocity***
- ***Deficits in nonverbal communicative behaviors used for social interaction***
- ***Deficits in developing and maintaining relationships and adjusting behavior to social contexts, appropriate to developmental level***

## 5. 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***

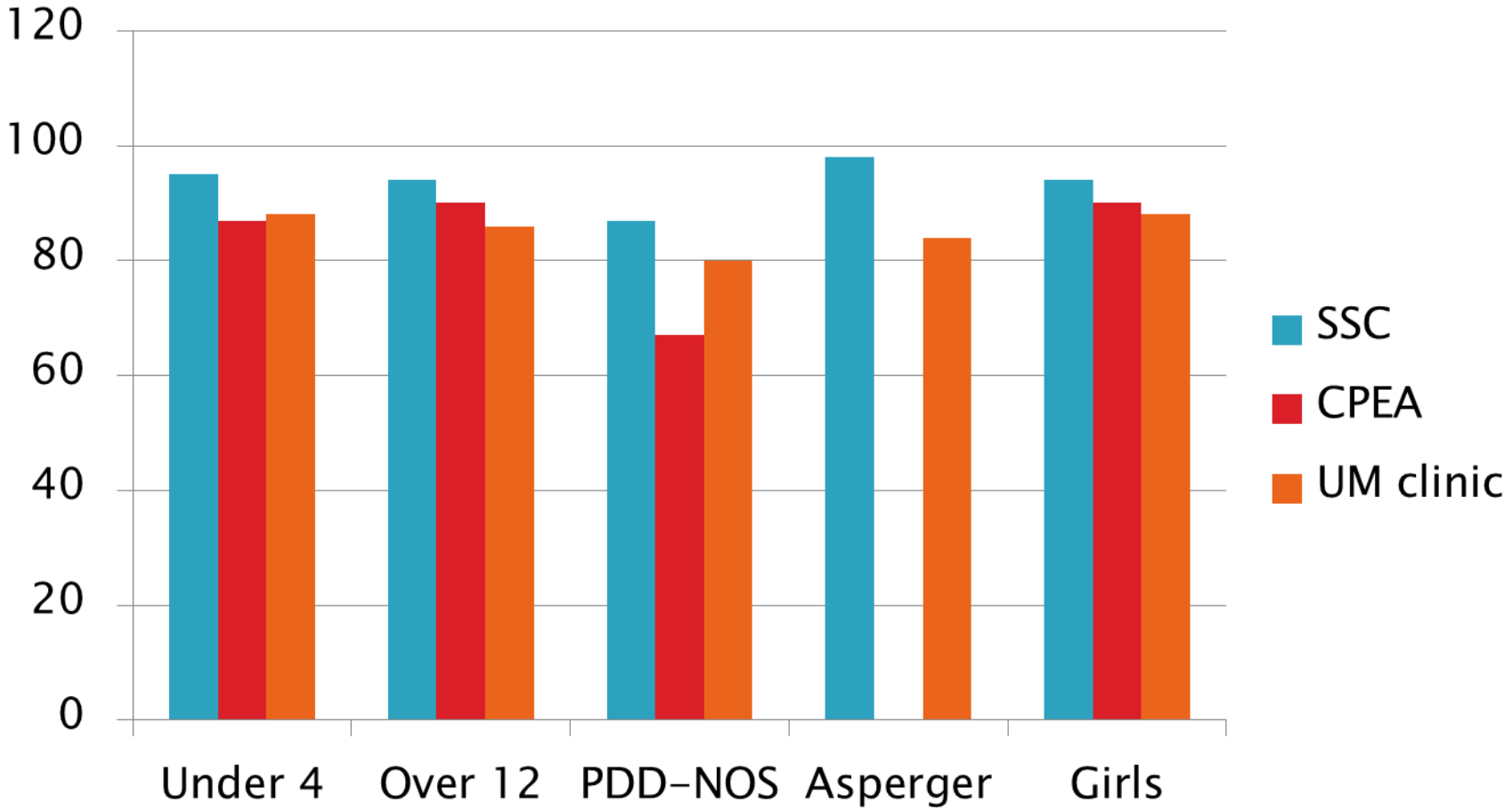
***D. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment***

# Sensitivity and Specificity of DSM-5 Criteria

| Study | ASD n | Sensitivity | Non-ASD n | Specificity |
|---|---|---|---|---|
| Frazier et al. (2012)[1] | 8911 | .96 | 5863 typ | .90 |
| McPartland et al. (2012)[2] | 657 | .61 | 276 non | .95 |
| Huerta et al. (2012)[3] | 1465 | .93 | 527 ref | .63 |

1. SRS and SCQ-L items mapped to DSM-5 criteria
2. DSM-IV checklist mapped to DSM-5 criteria
3. ADI-R items mapped to DSM-5 criteria

# Current sensitivities in special groups



6. Specifiers: With the new criteria, if the child has ASD symptoms, he or she gets an ASD diagnosis with a specifier for the etiology or associated medical condition:

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

Or

- ASD with tonic-clonic seizures
- ASD with chronic irritable bowel syndrome

# Important "time" issues

- Onset should be in early childhood
- DSM5 explicitly acknowledges that recognition is different than onset
- CAN'T have a clearly negative history into later childhood
- CURRENT IMPAIRMENT must be present though impairment is quantified by level of support needed

# 7. Early history is specified through:

Age of perceived onset

Pattern of onset (regression/no loss)

» Examples:

ASD with onset before 18 months and loss of words and social skills

ASD with onset by age 30 months and loss of social skills

ASD with no clear onset and no loss

# 8. Comorbidities can be formally acknowledged through additional diagnoses as specifiers:

- Intellectual disabilities
- Communication and language disorders
- Attention deficits and/or hyperactivity
- Mood disorders
- Oppositional behavior

| Dimensional Ratings for DSM 5 ASD | Social Communication | Fixated Interests and Repetitive Behaviors |
|---|---|---|
| Requires very substantial support | Minimal social communication | Marked interference in daily life |
| Requires substantial support | Marked deficits with limited initiations and reduced or atypical responses | Obvious to the casual observer and occur across context |
| Requires some support | Even with support, noticeable impairments | Significant interference in at least one context |
| | | |
| Subclinical symptoms | Some symptoms in this or both domains; no significant impairment | Unusual or excessive but no interference |
| | | |
| Normal variation | Maybe awkward or isolated but WNL | WNL for developmental level and no interference |

# *Pragmatic (Social) Communication Disorder (PSCD)*

1) is an impairment of pragmatics, only diagnosed when ASD is ruled out

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

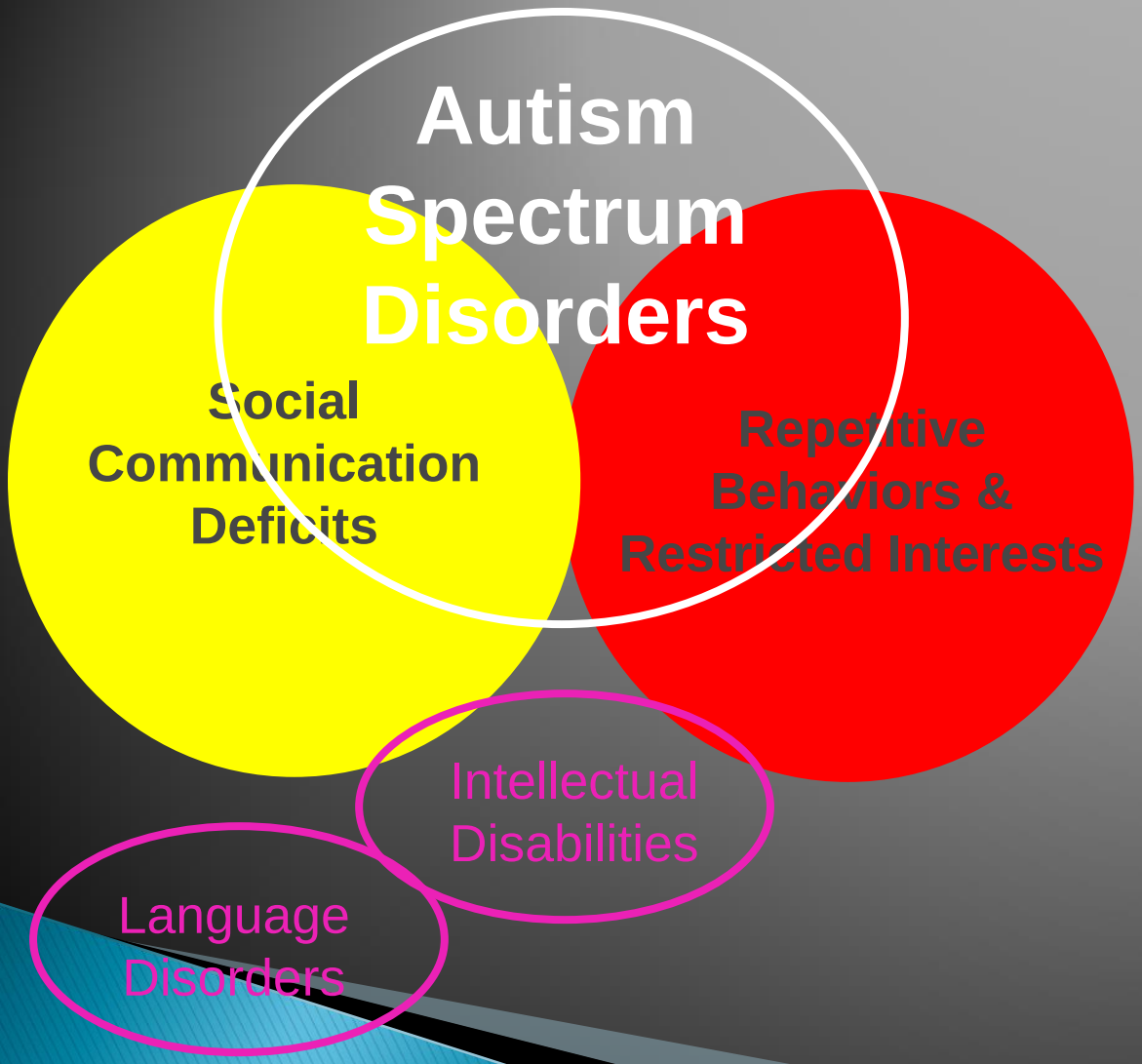
3) which affects the development of social relationships and discourse comprehension and

4) cannot be explained by low abilities in the domains of word structure and grammar or general cognitive ability

# Lingering questions

- Prioritizing “sensitivity” makes sense but has some dangers
- Diagnostic criteria for disorders are not the same as diagnoses
- Measuring outcomes and response to treatments (not same as dx)

# Autism Spectrum Disorders




- Sense of humor
- Fine motor skills
- Predictability
- Intelligence
- Visual-spatial skills
- Curiosity
- Attention to detail
- Honesty

# New opportunities on the horizon

- ADOS Severity scores by domain: social communication and repetitive behaviors
- Looking at combinations of ADOS and other measures (SRS, SCQ, new interview-ASI)
- Brief Observation of Social Communication (BOSC – formerly ADOS-C) for change, not diagnosis
- Observation of Spontaneous Expressive Language (OSEL) – measure of functional use of language for language levels of 2 to 5

# Autism is more than the sum of its parts

- Autism is not all that is problematic for many families and individuals (comorbidities including language delay, intellectual disabilities and other psychological disorders)
- Strengths and circumstances also make a difference – in the person with ASD and the family

# Collaborators and funding

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- Thanks to the children and families who have helped us in our studies

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