

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

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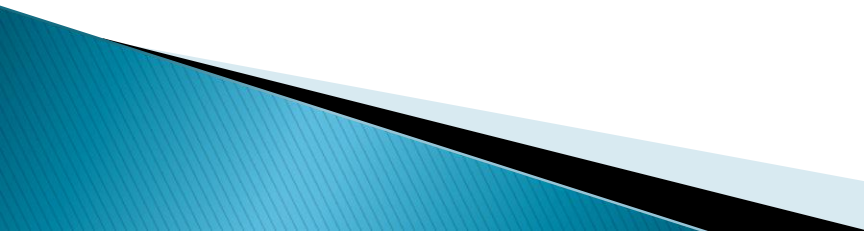
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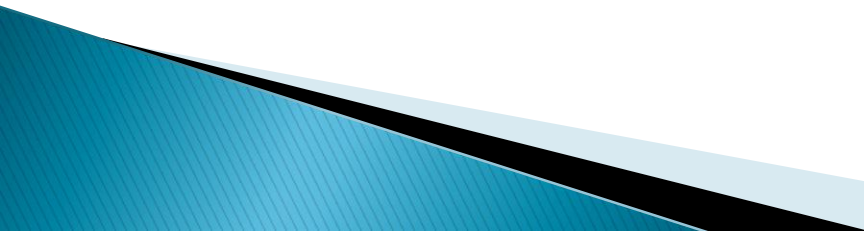
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\*Western Psychological Services

# DSM V Committee on Neurodevelopmental Disorders

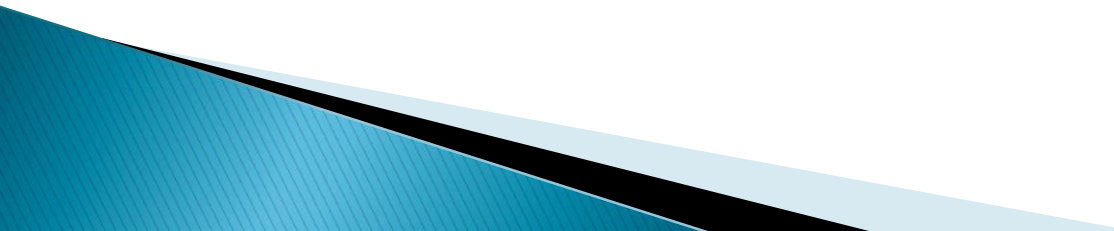
- ▣ Susan Swedo, M.D. , pediatrician and chair
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# Outline of talk

- ▶ 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

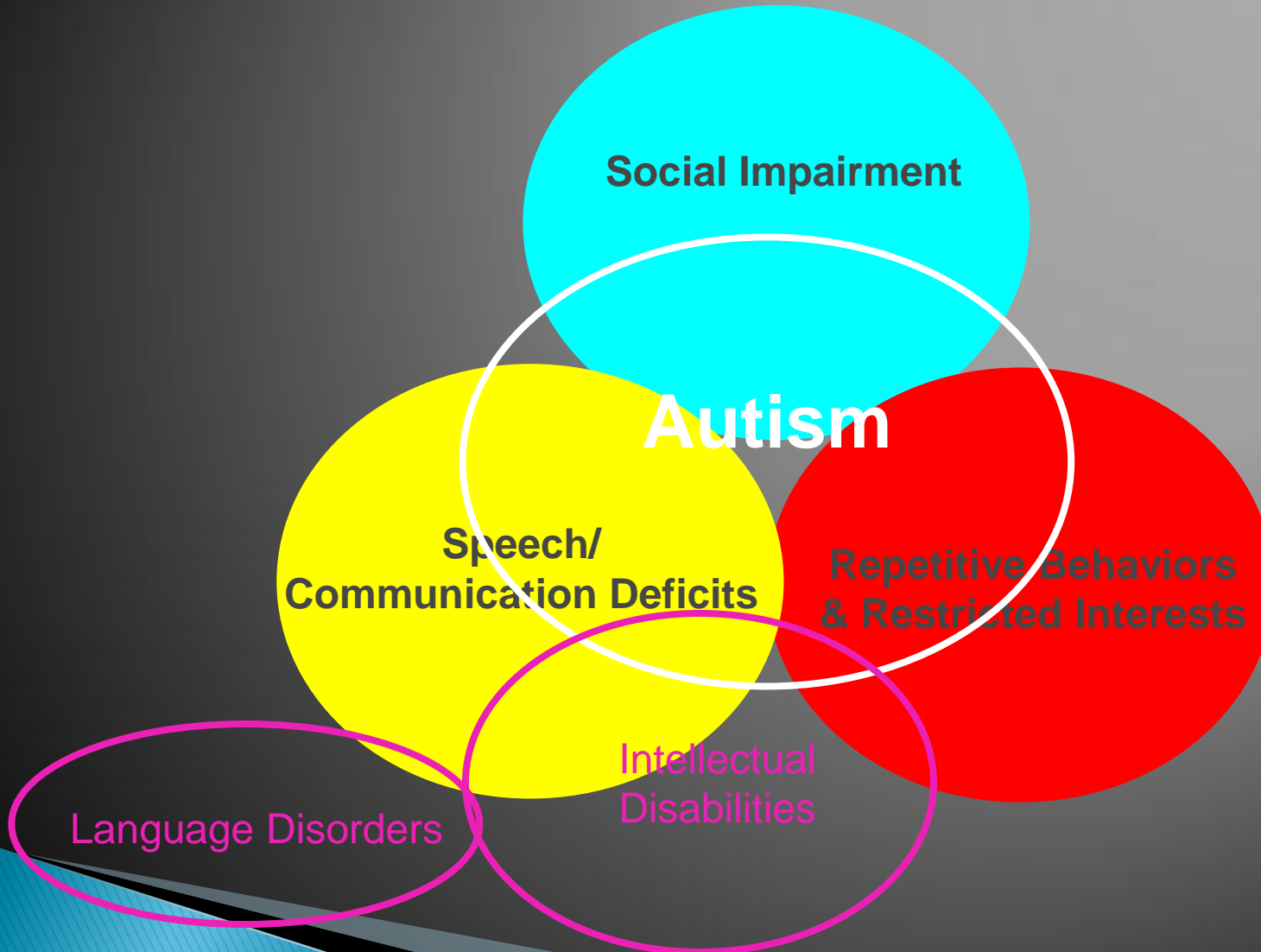
sk or



# Where are we now in diagnosis?

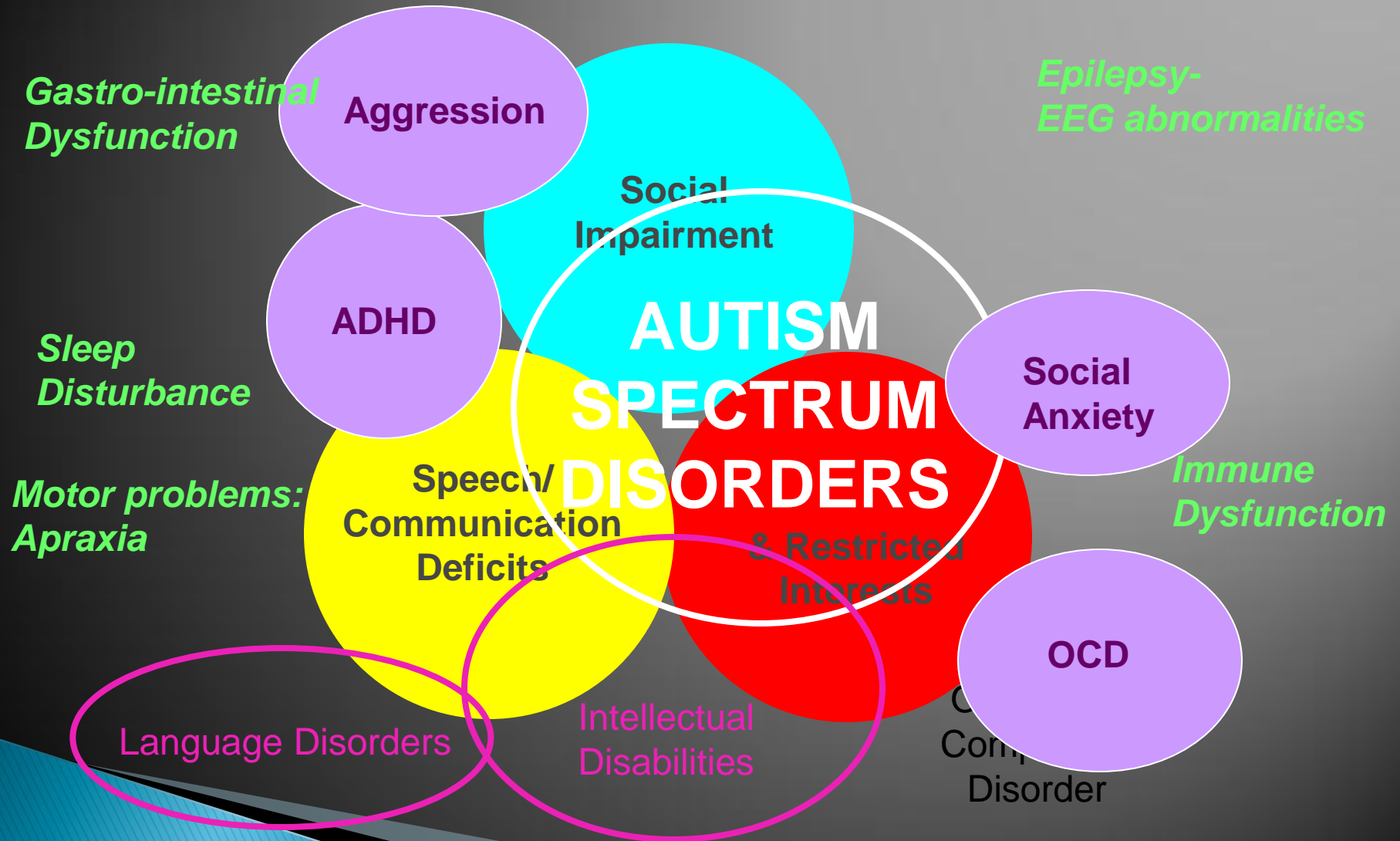
- Worldwide standard criteria (DSM IV/ICD-10)
- With **combined history**/informant report and **direct observation**, excellent sensitivity and specificity for prototypic autism in preschool and school age children
- Diagnoses of ASD are generally stable.
- Within a research program, clinical best estimates add to stability of a diagnosis.

# Pervasive Developmental Disorders



# Core Symptom Domains

PLUS Associated Medical Features (this does not count strengths, which would add other circles)



# 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?

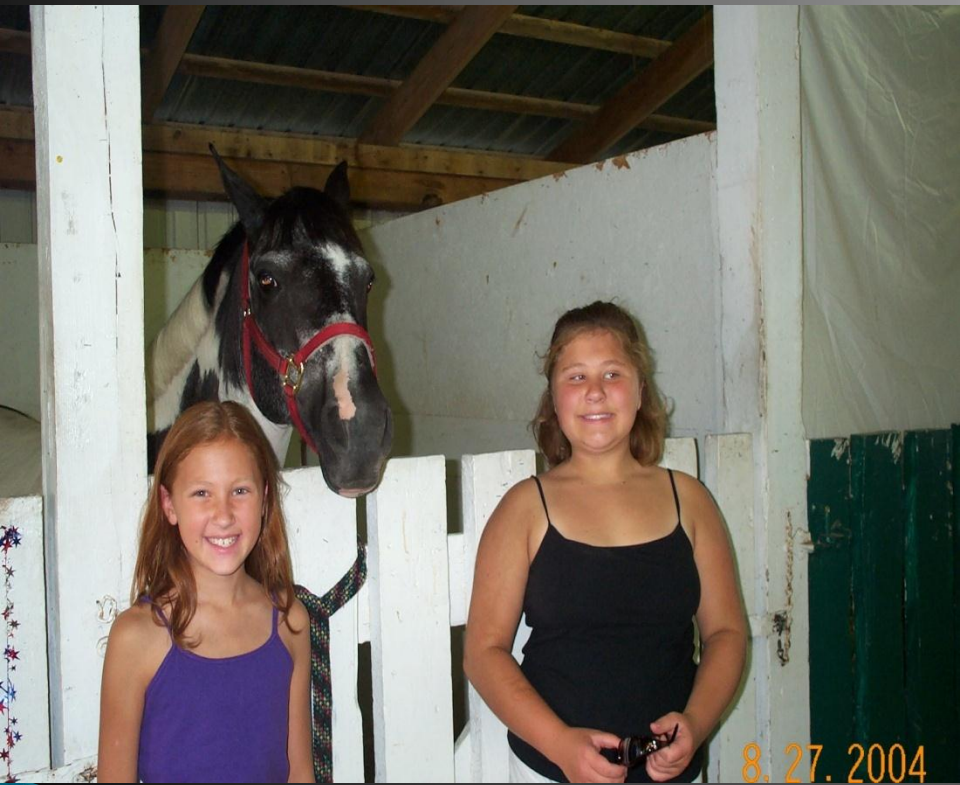
- ▶ Faster diagnoses = narrower comparisons.
- ▶ More specific diagnoses = age-related examples.
- ▶ Neurobiology = dimensions



# Draft Proposals for DSM V

- ▣ 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



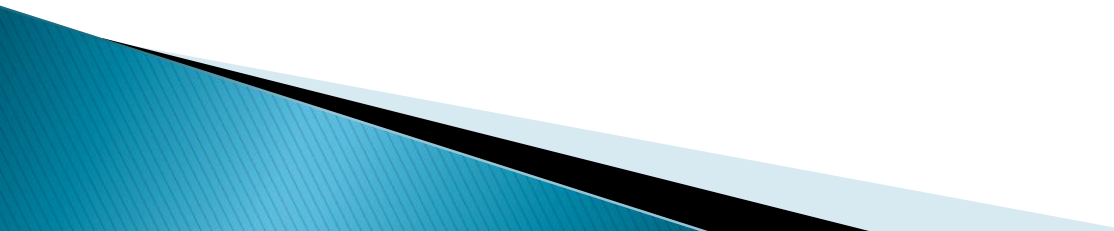
- ▶ Scientific validity
  - Questioning the importance of very early language milestones vs. fluent speech in older years
  - Overlap in research when VIQ controlled
- ▶ Concern about access to services



# 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](http://www.sfari.org) for interested scientists

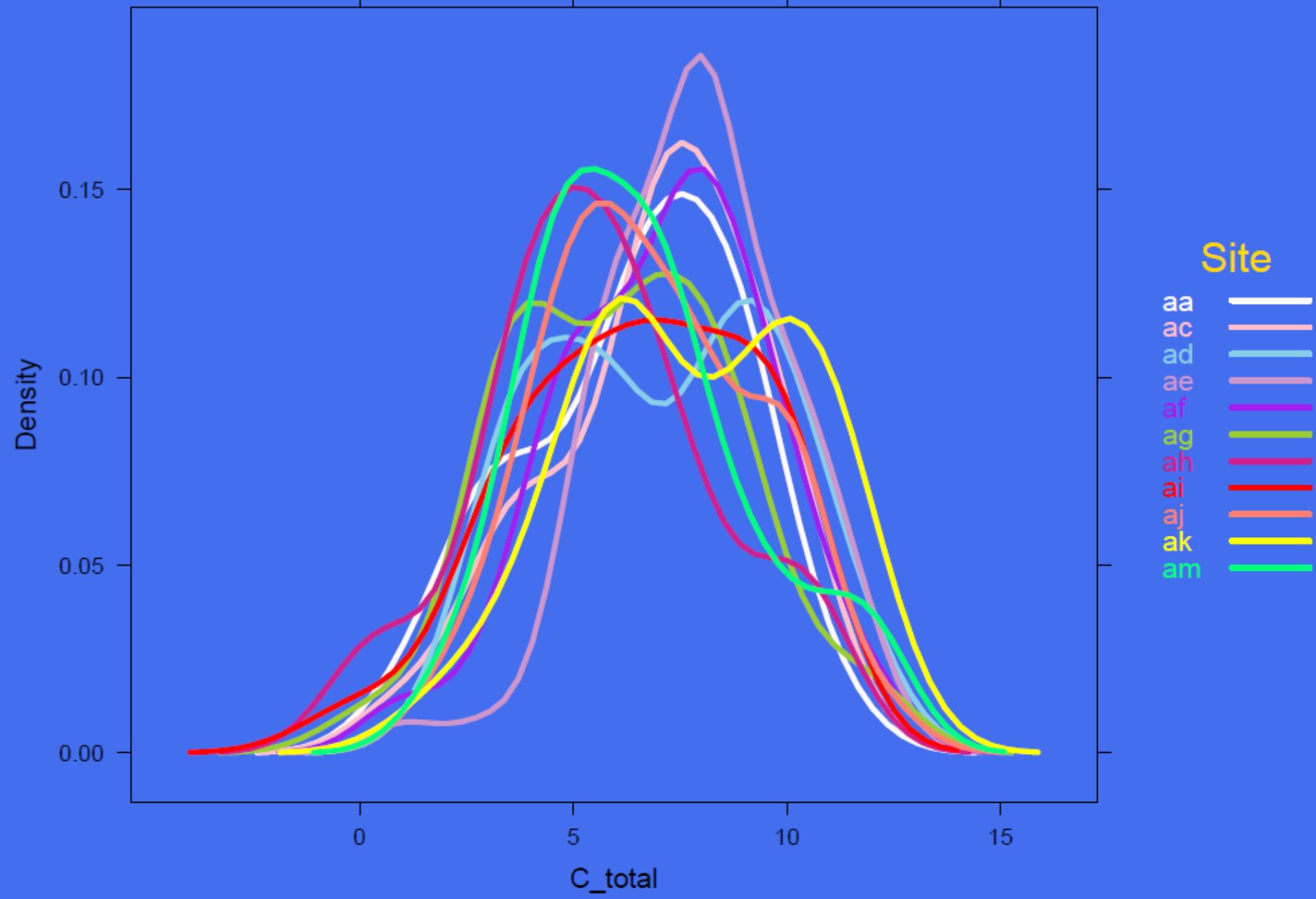
# Simons Simplex Collection

- ▶ 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
- 

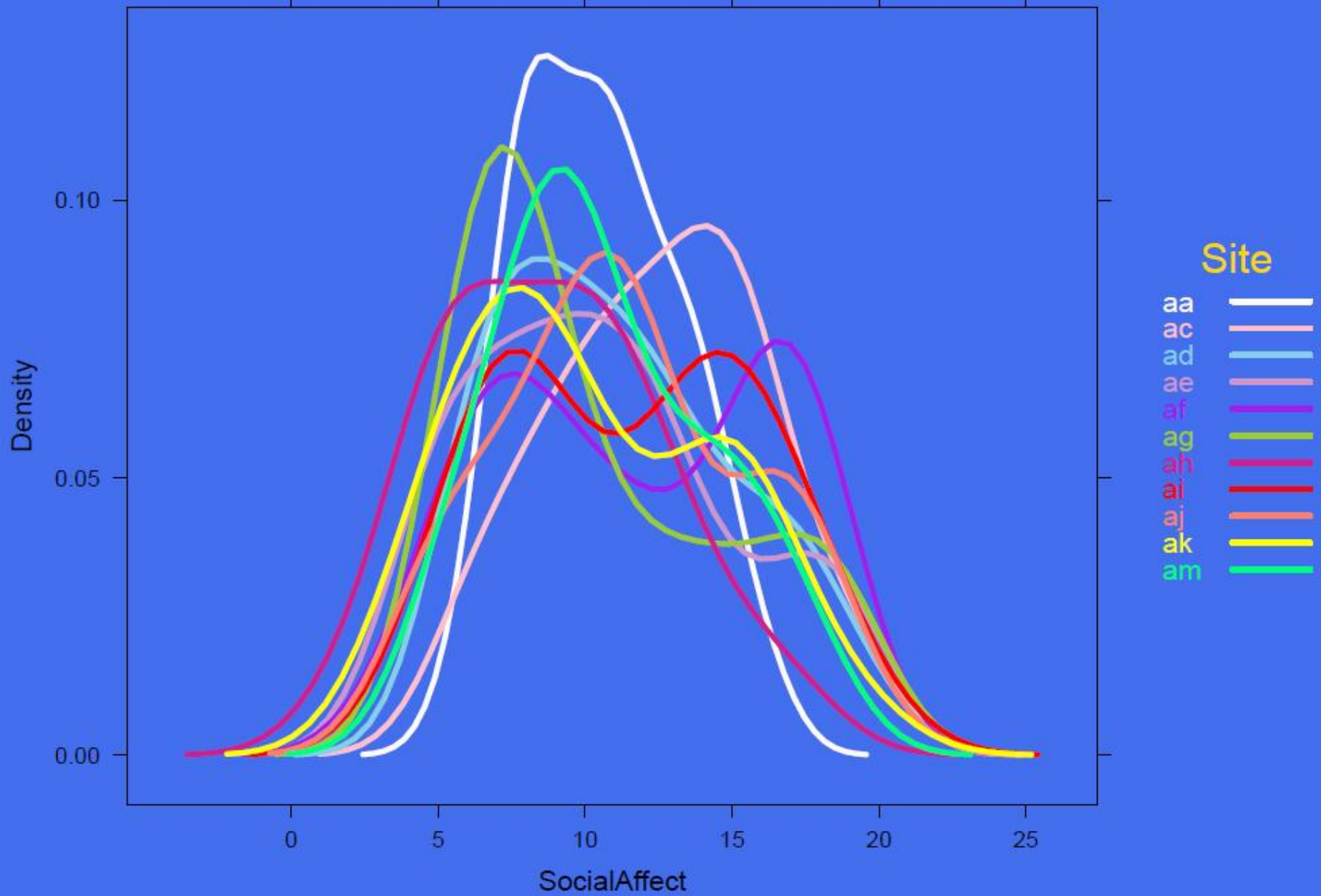
# Simons Simplex Collection



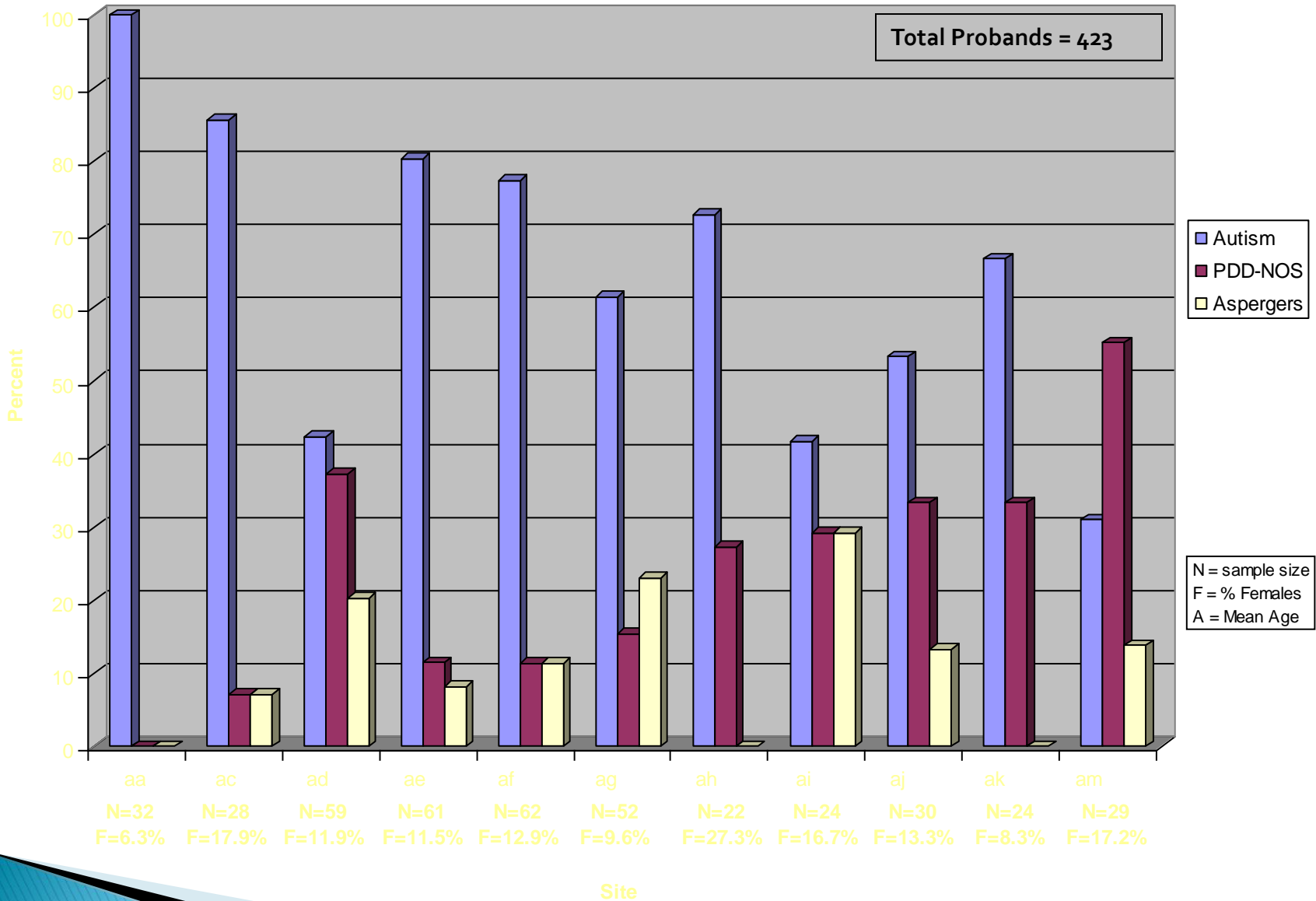
# ADI-R RRB Domain Scores

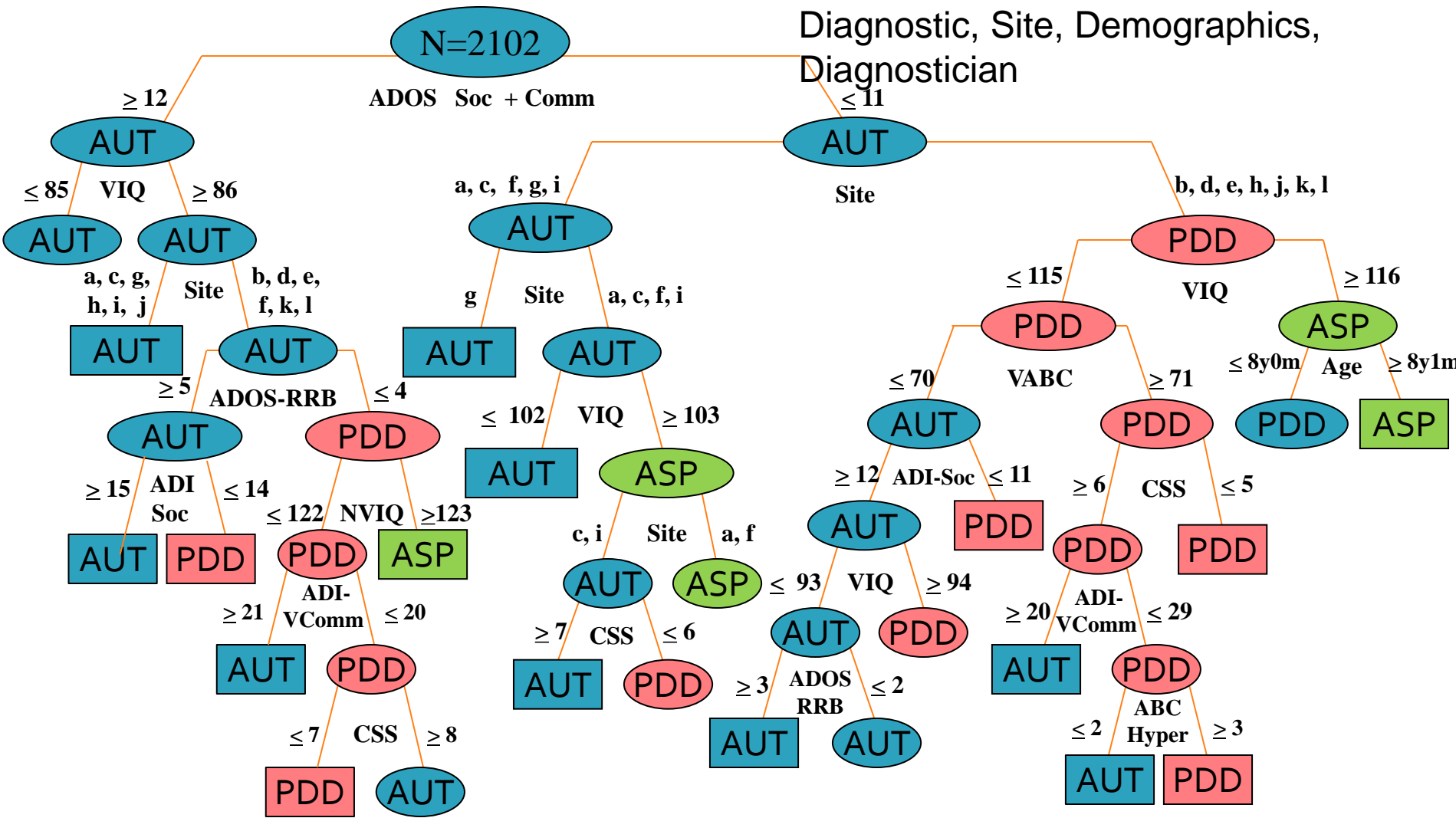


# ADOS Social Affect



# ASD Distribution of Probands





*The Simons Simplex Collection*

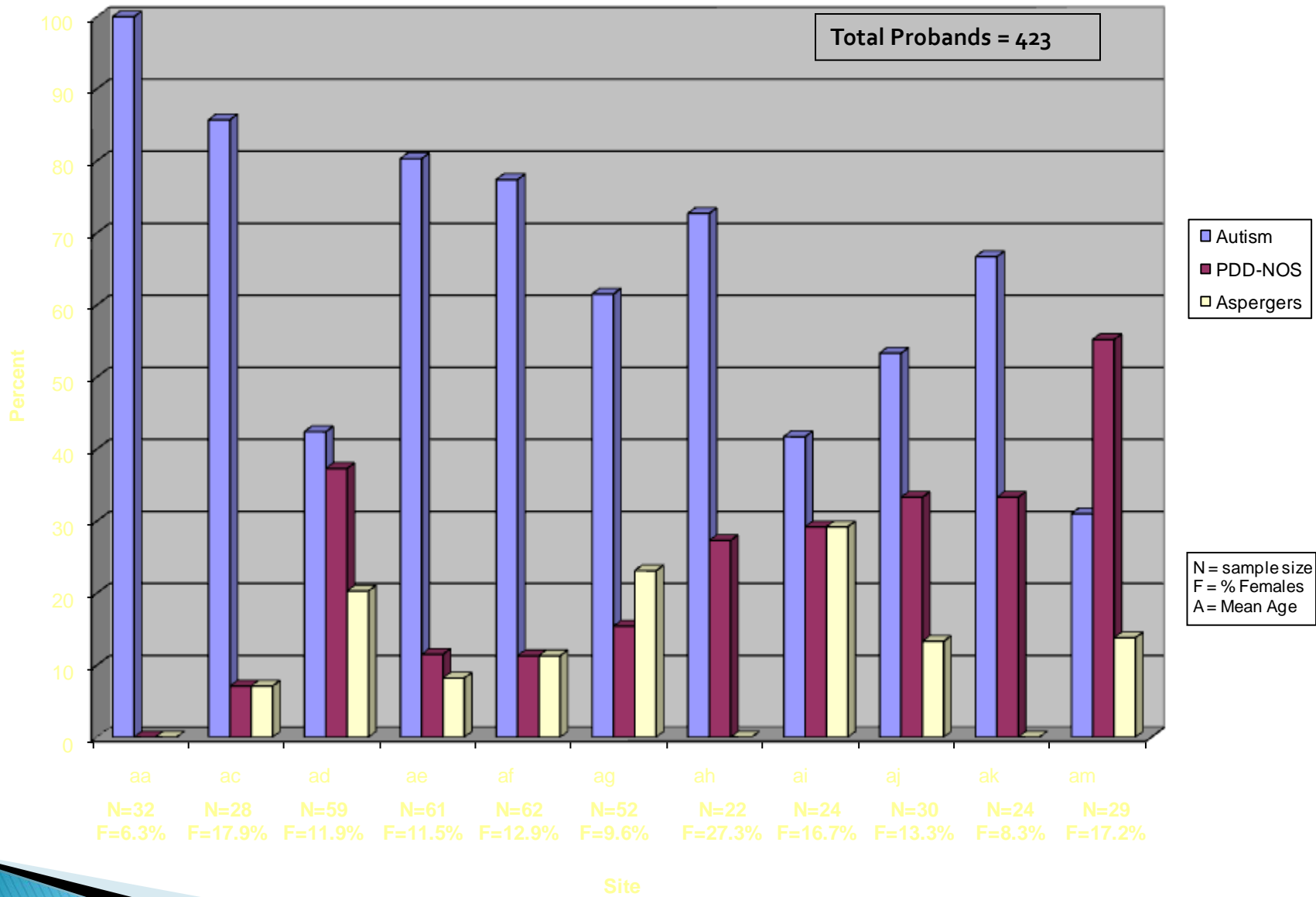
# Predictors of various ASD diagnoses by site

	a	b	c	d	e	f	h	i	l	k
1 <sup>st</sup> 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
2 <sup>nd</sup> 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



# ASD Distribution of Probands



# 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



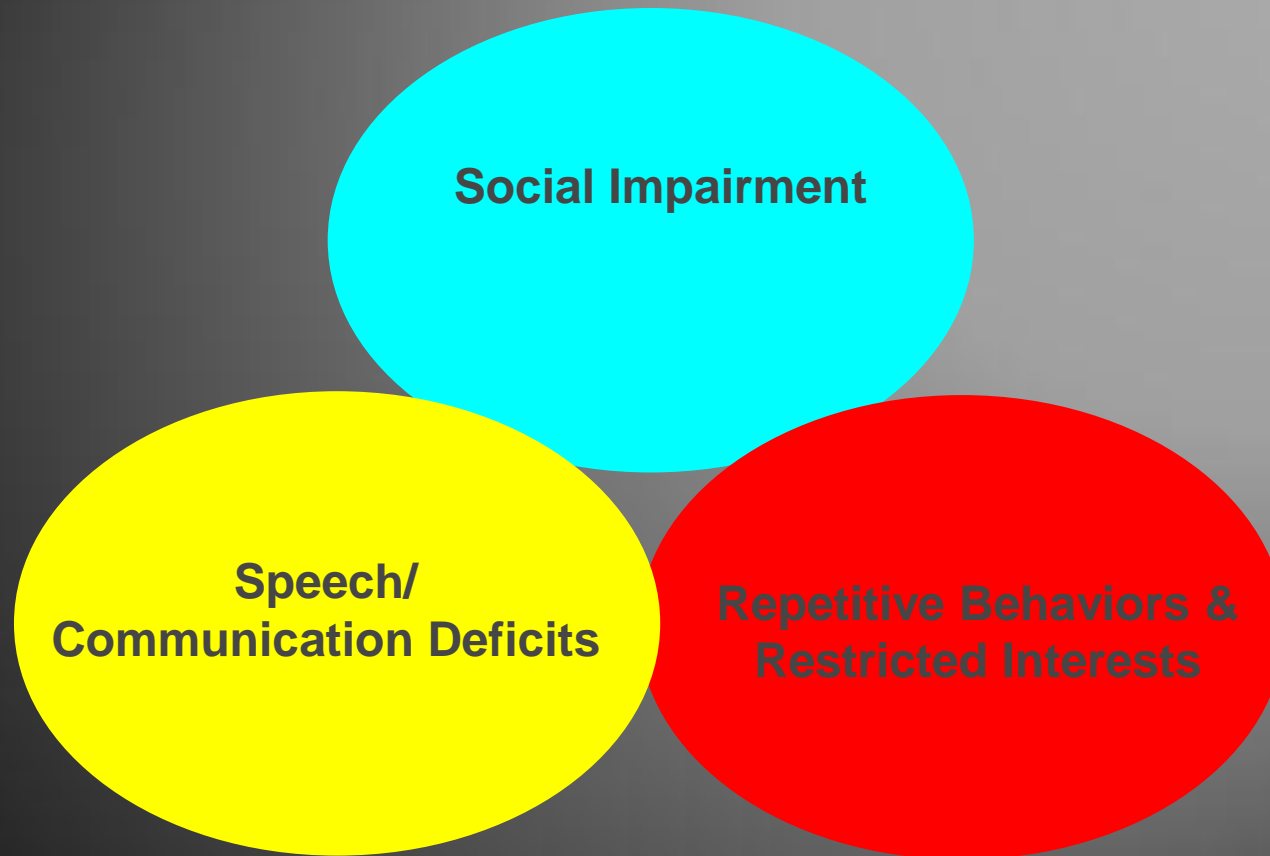
## 2. How many domains?

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

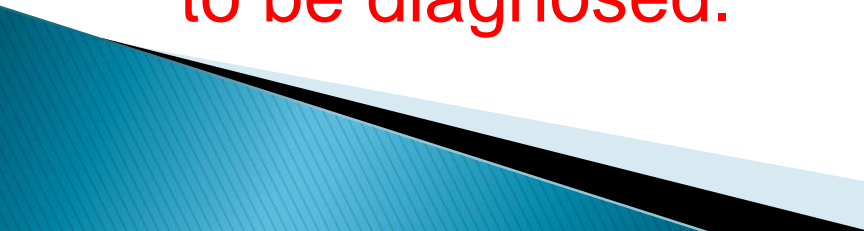


# ***Social Communication Disorder (SCD)***

- 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 >>>

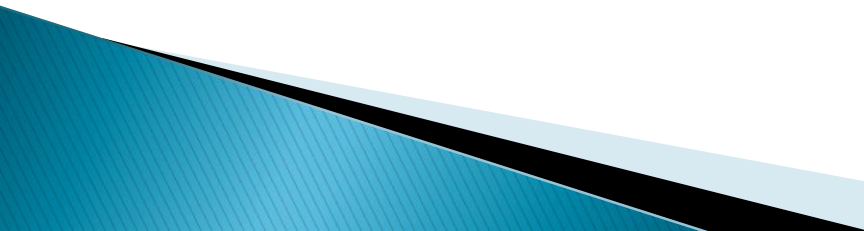
Or a modifier indicating another important factor:

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)

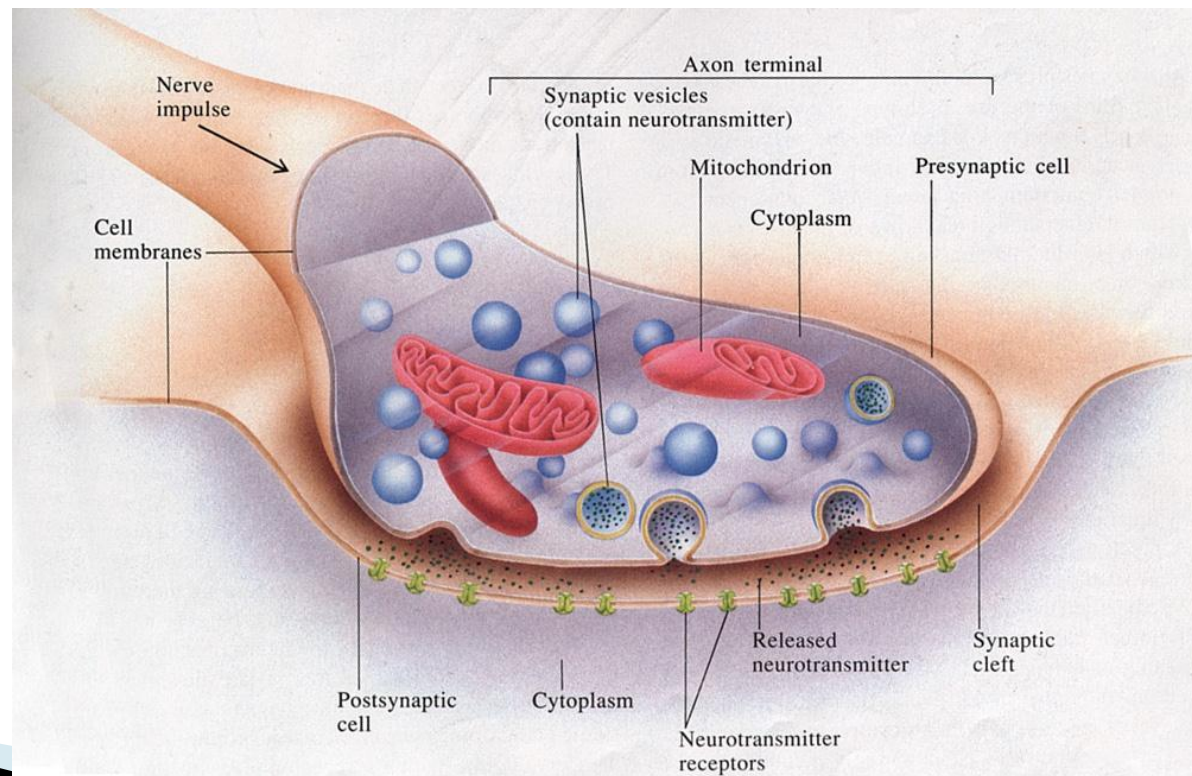
- ▶ For a variety of domains relevant to almost any psychiatric condition
  - ▶ Some of them are:
    - Developmental level or nonverbal and verbal IQ
    - Adaptive functioning
    - Verbal abilities at the time of intake
    - Hyperactivity/impulsivity
    - Sleeping difficulties
    - Co-occurring 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

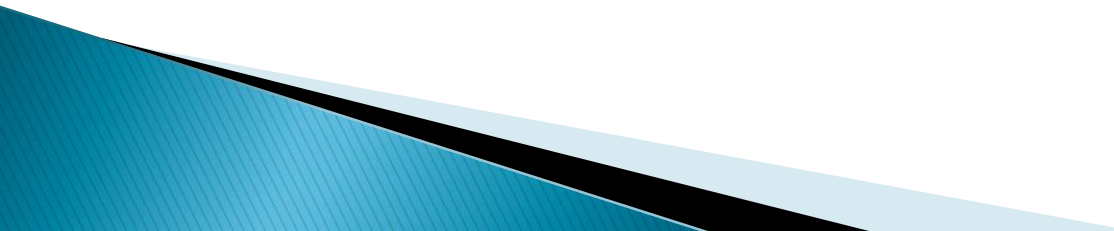
Dimensional Ratings for DSM5 ASD	Social Communication	Fixated Interests and Repetitive Behaviors
Requires very substantial support	Minimal social communication	Marked interference in daily life
Requires substantial support	Marked deficits with limited initiations and reduced or atypical responses	Obvious to the casual observer and occur across context
Requiring support	Without support, some significant deficits in social communication	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

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 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)



# 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.

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