

Autism Spectrum Disorder: Overview of Diagnosis and Assessment

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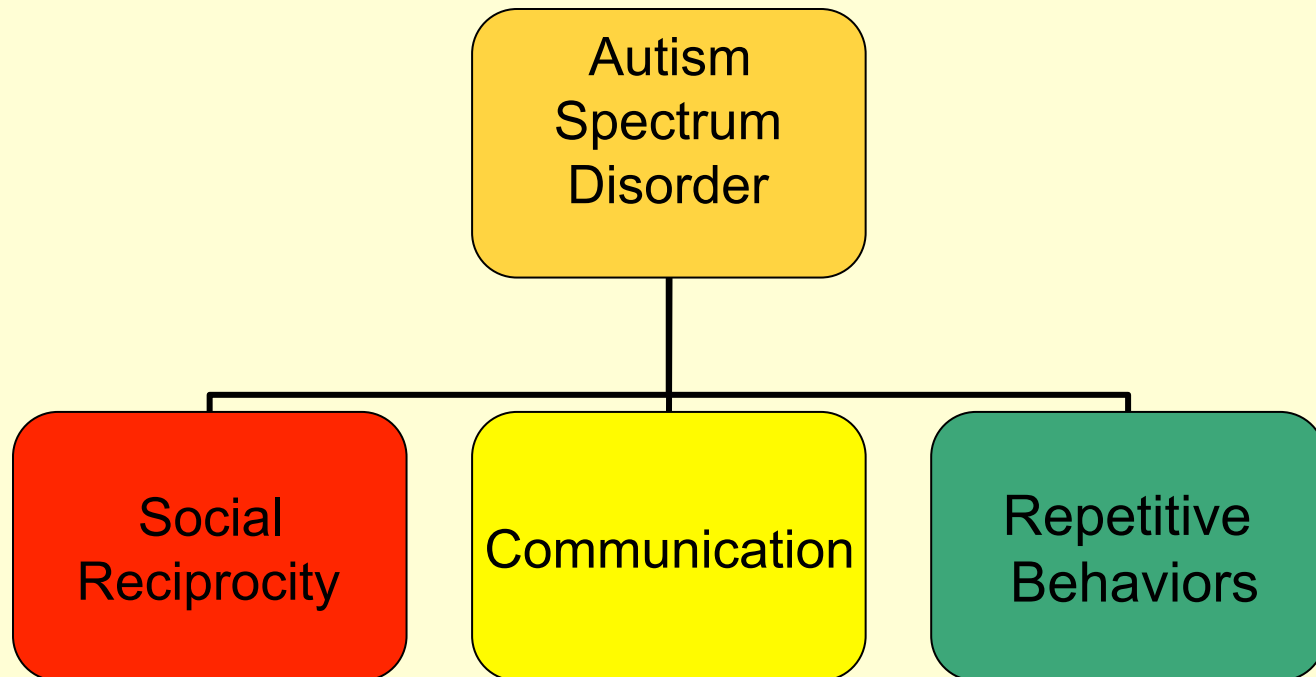
What Is Autism Spectrum Disorder?

Developmental disorder

Affects communication, reciprocal social interactions and play, interests, and behavior

Symptoms are present prior to 3 years of age

It affects development and is affected by development



Demographics of ASD

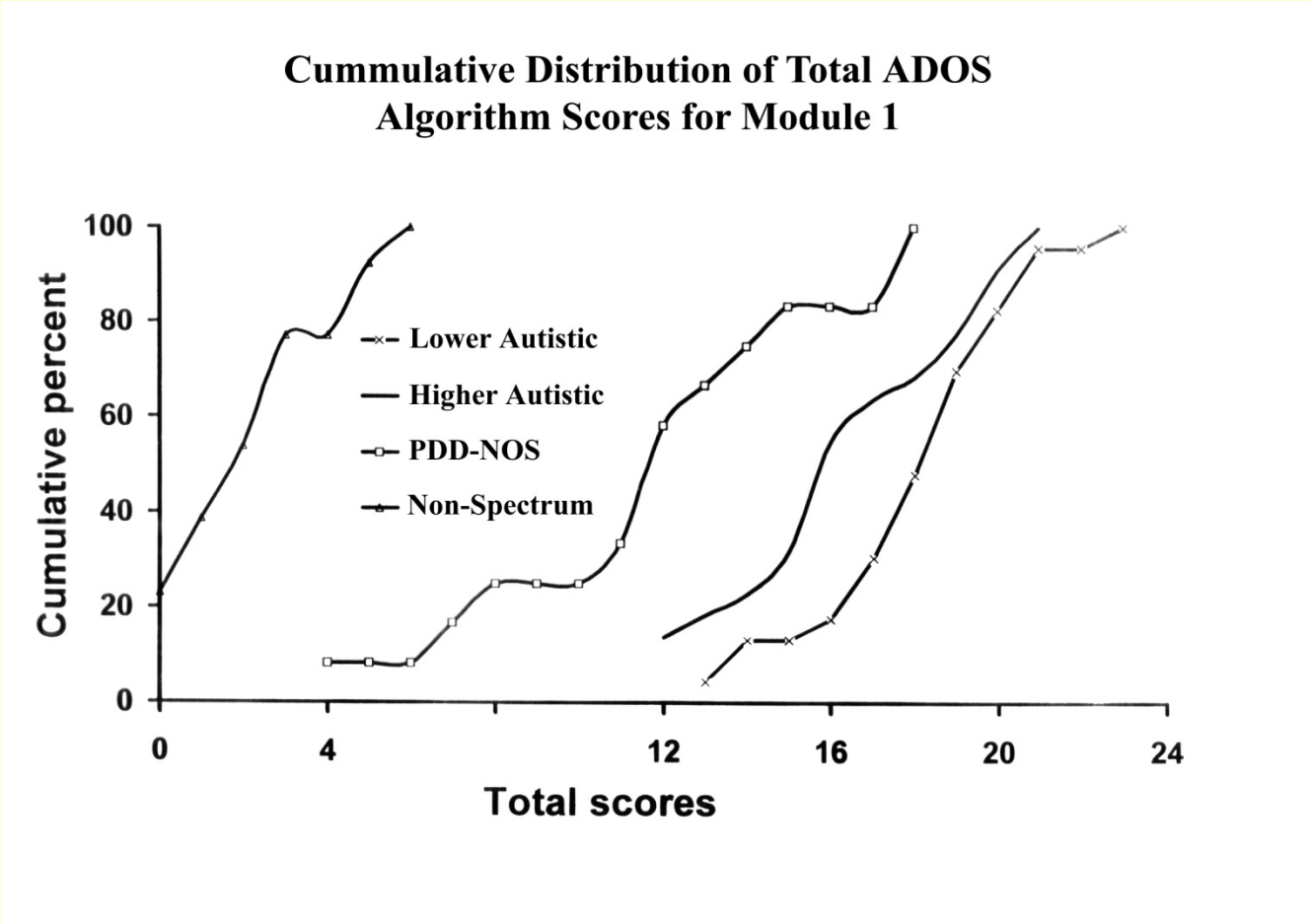
- Prevalence of autism spectrum disorders is consistent around the globe
- Four times more prevalent in boys than in girls.
- No racial, ethnic, social boundaries
- Not related to family income, lifestyle, or educational levels
- Present before the age of 3, but diagnosis often later
- Historically, 70–85% of cases of autism were associated with intellectual disability.
 - Current estimate is 50% will have intellectual disability
- Similarly, the statistic that half of all children with autism will not speak is often cited, but recent estimates suggest that 75–90% of children with ASD have some functional language.

Current, Categorical Definition

- Pervasive Developmental Disorders (PDD)
- PDD = an “umbrella category”
- Autism
- Atypical Autism (PDD–NOS)
- Asperger’s Syndrome
- Childhood Disintegrative Disorder
- Rett’s Syndrome



Children with ASDs do not fall easily into separable categories



ASD Defies Generalization

Measured Cognitive Skills

Severe-----Gifted

Social Interaction

Aloof-----Passive-----Active but odd

Communication

Nonverbal-----Verbal

Behaviors

Intense-----Mild

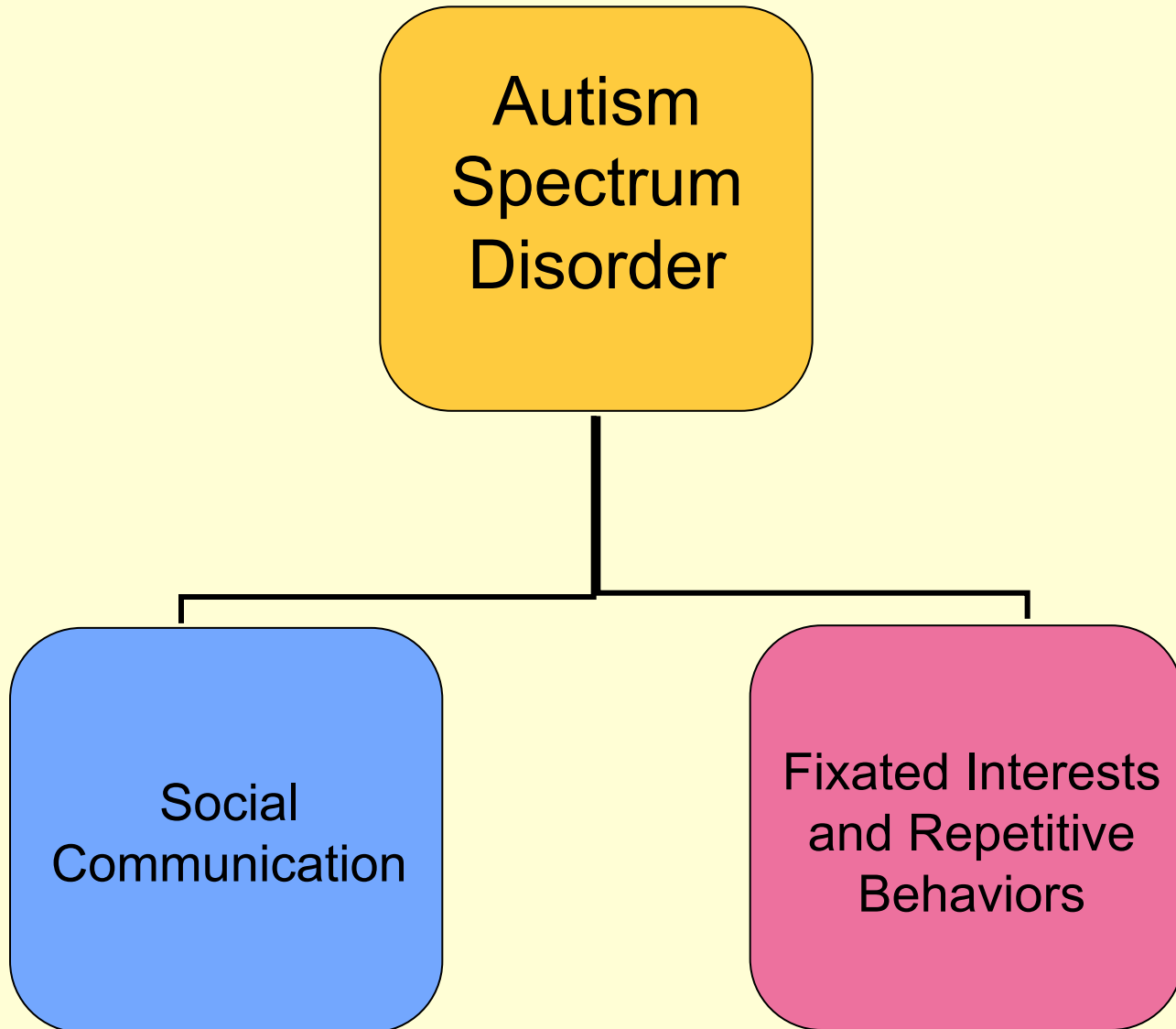
Sensory

Hyposensitive-----Hypersensitive

Motor

Uncoordinated-----Coordinated

DSM-V Proposed Revisions



Rationale for DSM-V

- Changes made to better reflect state of knowledge about pathology and clinical presentation
- Distinctions among ASD “subtypes” have been found to be inconsistent over time, variable across sites, and often associated with severity, language level, or intelligence rather than features of the disorder.
- Because ASD is defined by a common set of behaviors, it is best represented as a single diagnostic category that is adapted to the individual’s clinical presentation by inclusion of clinical specifiers (e.g., severity, verbal abilities and others) and associated features (e.g., known genetic disorders, epilepsy, intellectual disability and others).

(video clips)

DSM-IV: **Social** (need 2), **Communication** (need 1)

- a. Marked impairment in the use of **multiple nonverbal behaviors** such as eye-to-eye gaze, facial expression, body postures, and gestures to regulate social interaction
- b. Failure to develop **peer relationships** appropriate to developmental level
- c. Lack of **spontaneous seeking to share** enjoyment, interests, or achievements with other people (e.g., by lack of showing, bringing, or pointing out objects of interest)
- d. Lack of **social or emotional reciprocity**
- e. Delay in, or total lack of, the **development of spoken language** (not accompanied by an attempt to compensate through alternative modes of communication such as gesture or mime)
- f. In individuals with adequate speech, marked impairment in the ability to **initiate or sustain a conversation** with others
- g. **Stereotyped and repetitive** use of language or idiosyncratic language
- h. Lack of varied, spontaneous **make-believe play or social imitative play** appropriate to developmental level

DSM-V (need all)

- 1. Deficits in **social-emotional reciprocity**; ranging from abnormal social approach and failure of normal back and forth in 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 and maintaining **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 people.

DSM–IV RRB (1)

- a. Encompassing preoccupation with one or more stereotyped and **restricted patterns of interest** that is abnormal either in intensity or focus
- b. Apparently **inflexible adherence** to specific, nonfunctional **routines** or rituals
- c. Stereotyped and repetitive **motor mannerisms** (e.g., hand or finger flapping or twisting, or complex whole–body movements)
- d. Persistent **preoccupation with parts** of objects

DSM–V RRB (2)

1. Stereotyped or **repetitive speech, motor movements, or use of objects**; (such as simple motor stereotypies, echolalia, repetitive use of objects, or idiosyncratic phrases).
2. Excessive **adherence to routines**, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change; (such as motoric rituals, insistence on same route or food, repetitive questioning or extreme distress at small changes).
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 spinning objects).

Prevalence of RRBs across age and NVIQ

(Bishop, Richler, & Lord, 2006)

High-frequency	Low-frequency	Highly Variable
Unusual sensory interests	Unusual preoccupations	Circumscribed interests
Repetitive use of objects	Abnormal idiosyncratic responses to sensory	Compulsions and rituals
Stereotyped speech	Unusual attachments	Sensitivity to noise
	Resistance to change in environment	Difficulties with change in routine
	Self-injury	Hand and finger mannerisms
	Verbal rituals	Other complex mannerisms

RRBs vary with time and NVIQ

(Bishop, Richler, & Lord, 2006)

- As age increased, the relationship between RRBs and NVIQ increased.
 - Relationship generally negative, *except Circ. Ints. and Rituals*
- In children under 3, little relationship between RRBs and NVIQ.
- For some RRBs, children with lower NVIQs tended to have more *severe* behaviors

Diagnostic measures

- Goal: classification
 - Reliable across time
 - Diagnosis reflects core symptoms
 - Relationship to gold standards/validity
- Provide information about associated features, response to treatment, prognosis, and etiology.
- Provide access to services and support.



Diagnostic Evaluation

- Necessary Ingredients:
- Developmental History Interview
- Structured observation including direct interaction with child
- Establish developmental levels—needed for differential diagnosis
 - Cognitive testing (separate nonverbal and verbal IQ scores)
 - Communication testing (separate expressive and receptive scores)
 - Adaptive skills: children with ASD usually score much lower on Adaptive skills than is expected based on cognitive scores
- Deviance v. Delay:
- Uneven development supports ASD
- Vocabulary and cognitive skills may be average or better; use of social communication is odd or atypical.
- Even with delays in development, social skills are significantly below expectations

- Both positive (abnormal) behaviors, and negative (the absence of normal) behaviors are required to make a diagnosis of ASD.
- This means that developmental level and contextual effects (in what kind of circumstances does the child or adult function?) can both have significant effects on diagnostic judgments.



Measures often used in diagnostic evaluations for ASD

• **Autism diagnostic measures:**

- Autism Diagnostic Interview-Revised
- Autism Diagnostic Observation Schedule (ADOS)

• **Checklists & Screeners:**

- Social Responsiveness Scale
- Social Communication Questionnaire
- (Under 2) Communication and Symbolic Behavior Scales
- (Under 3) Modified Checklist for Autism in Toddlers (M-CHAT)
- Childhood Autism Rating Scale (CARS)

Other measures:

- Adaptive: Vineland-II
- Cognitive
 - Differential Abilities Scales (DAS-2)
 - Mullen Scales of Early Development
- Communication
 - CELF-4, CELF-P2
 - Sequenced Inventory of Communication Development (SICD)
 - Comprehensive Assessment of Spoken Language (CASL)
 - PPVT-4
 - Preschool Language Scale (PLS-4)
 - MacArthur Communication Development Inventories

The Autism Diagnostic Interview, Revised

- Rutter, Le Couteur, Lord (2003)

Investigator-based vs. Respondent-based Interviews (semi-structured vs. structured)

	<i>Investigator-based</i>	<i>Respondent-based</i>
CONCEPT	investigator's	subject's
STANDARDIZATION	a) coverage b) concept	coverage wording of probes
STYLE	conversational	questionnaire
TYPE of QUESTIONS	mainly open	closed
TIMING	use of personalized anchor-points	use of calendar dating
SCOPE OF CODING	all behavioral descriptions	answers only to specified questions
WHAT is CODED	Investigator's analysis of behavioral descriptions	subject's "yes"/ "no" to closed questions

Organization of ADI-R

1. General orientation
2. Early developmental history
3. Communication and language
4. Social development and play
5. Unusual interests and behaviors
6. Various nonspecific behaviors plus special skills

Example of Criteria:

Item 34: Social Vocalization/Chat

REQUIRES: Reciprocity
 Turn Taking
 Maintenance of Interchange
 Social Quality

Scene Setting Opening:

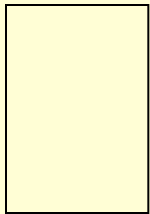
“When children are babbling or just beginning to talk, they sometimes seem to be making sounds just to be friendly and sociable, rather than because they want something.”

“...often they chatter away, following their parents around even though they know only a few words.”

Description/ Example Required:

Sample ADI-R communication code:

34. Social Verbalization/Chat:



0 = verbalizes or chats with clear social quality of talking to be friendly or to express interest, rather than to make needs known

1 = some social use of speech in response to caregiver or to get attention with no other obvious motivation, but limited in frequency or range of contexts

2 = uses some speech to alert caregiver to immediate needs or wants, but little or no purely social use of verbalization

8 = N/A

9 = N/K or not asked

Autism Diagnostic Observation Schedule

- Lord, Rutter, DiLavore, Risi (2002)

The ADOS creates a “social world” in which behaviors related to the autism spectrum can be observed:

- Structured and unstructured activities
 - Young ages/lower language level: mixture of structured and unstructured play, imitation, to structured beginning language activities
 - Higher language level/age: structured activities as well as informal conversation, interview questions about daily living, relationships, and emotional understanding
- Guidelines for “hierarchy” of examiner’s behavior
- Dependent on examiner’s experience and sensitivity (to act and not to act)

The ADOS is standardized by:

- Tasks and activities
- Materials
- Behavior of the examiner
- Behaviors to be observed
- How the individual's behaviors are quantified
- How the diagnosis is achieved
- Training of examiner
- Standards for achieving and maintaining reliability

Sensitivities/Specificities Autism

Risi et al., 2006

Diagnostic Criteria		True Positives (n)	True Negatives (n) ^a	False Positives (n) ^b	False Negatives (n)	Sensitivity (95% CI) ^c	Specificity (95% CI) ^c
ADI-R and ADOS (AUT)	3+	443	361 (204)	59 (53)	97	82.0%	86.0%
	<36	131	94 (53)	14 (12)	31	80.9%	87.0%
	ID	41	11 (6)	11 (6)	4	91.1%	50.0%
ADOS (AUT)	3+	497	309 (161)	111 (96)	43	92.0%	73.6%
	<36	158	64 (28)	44 (37)	4	97.5%	59.3%
	ID	45	4 (2)	18 (10)	0	100%	18.2%

Sensitivities / Specificities PDD

Diagnostic Criteria		True Positives (n)	True Negatives (n) ^a	False Positives (n) ^b	False Negatives (n)	Sensitivity (95% CI) ^c	Specificity (95% CI) ^c
ADI-R* and ADOS (ASD)	3+	157	143	20	100	61.1%	87.7%
	<36	54	34	9	11	83.1%	79.1%
	ID	10	2	8	2	83.3%	20.0%
ADOS (ASD)	3+	186	127	36	71	72.4%	77.9%
	<36	63	29	14	2	96.9%	67.4%
	ID	11	1	9	1	91.7%	10.0%

ADOS UMN Clinic

Final Diagnosis	ADOS Classification		
	Autism N, %	ASD N, %	Nonspectrum N, %
Autism (N=47)	46 , 98%	0, 0%	1, 2%
PDD/Asperger (N=15)	6, 40%	5, 33%	2, 13%
Nonspectrum (N=19)	5, 26%	1, 0.5%	13, 68%

Sensitivity and Specificity

ADOS Classification	Best Estimate Diagnosis:		
	Autism v. Nonspectrum	Any ASD v. Nonspectrum	Non-autism ASD v. Nonspectrum
Autism			
Sensitivity	0.98	0.84	0.40
Specificity	0.74	0.74	0.74
ASD			
Sensitivity	0.98	0.97	0.73
Specificity	0.68	0.68	0.68

Core symptoms and Related conditions

- Core symptoms define the diagnosis, are represented in diagnostic criteria
- Related behaviors are discussed in DSM text descriptions

ASD Core symptoms:

Social interaction
Qualitative impairment in
communication
Repetitive behaviors and
restricted interests

ASD related behaviors:

Intellectual disability
Speech/language delays
Adaptive skill deficits
Attention problems
Hyperactivity/impulsivity
Aggressive behaviors
Anxiety/depression
Seizures
Sleep problems

Related conditions common to ASD: The CBCL

J Psychopathol Behav Assess (2011) 33:31–37

Table 3 Percent of children at or above the borderline clinical range for the CBCL scales ($t \geq 67$)

CBCL scale	ASD samples %	Control sample %
Total score	63.1	1.5
Anxious/depressed	48.3	0.0
Withdrawn/depressed	55.5	3.0
Somatic complaints	19.5	4.5
Social problems	71.2	1.5
Thought problems	82.4	4.5
Attention problems	64.1	1.5
Rule-breaking behavior	10.2	0.0
Aggressive behavior	22.0	1.5

Differential Diagnosis

Discriminating ASD from other disorders is easiest for school age children with some language, who are not fluent speakers. As we move up and down the age span, and up and down levels of mental retardation and language delay, discriminations become more difficult.

Deviance v. Delay:

Uneven development supports ASD

Vocabulary and cognitive skills may be average or better; use of social communication is odd or atypical.

Even with delays in development, social skills are significantly below expectations

Clarifying ASD and Intellectual Disability

- A good developmental history, with specific focus on patterns indicative of autism, is important.
- Focus on social behaviors, social directedness of communication
- Repetitive sensorimotor behaviors are not as reliable

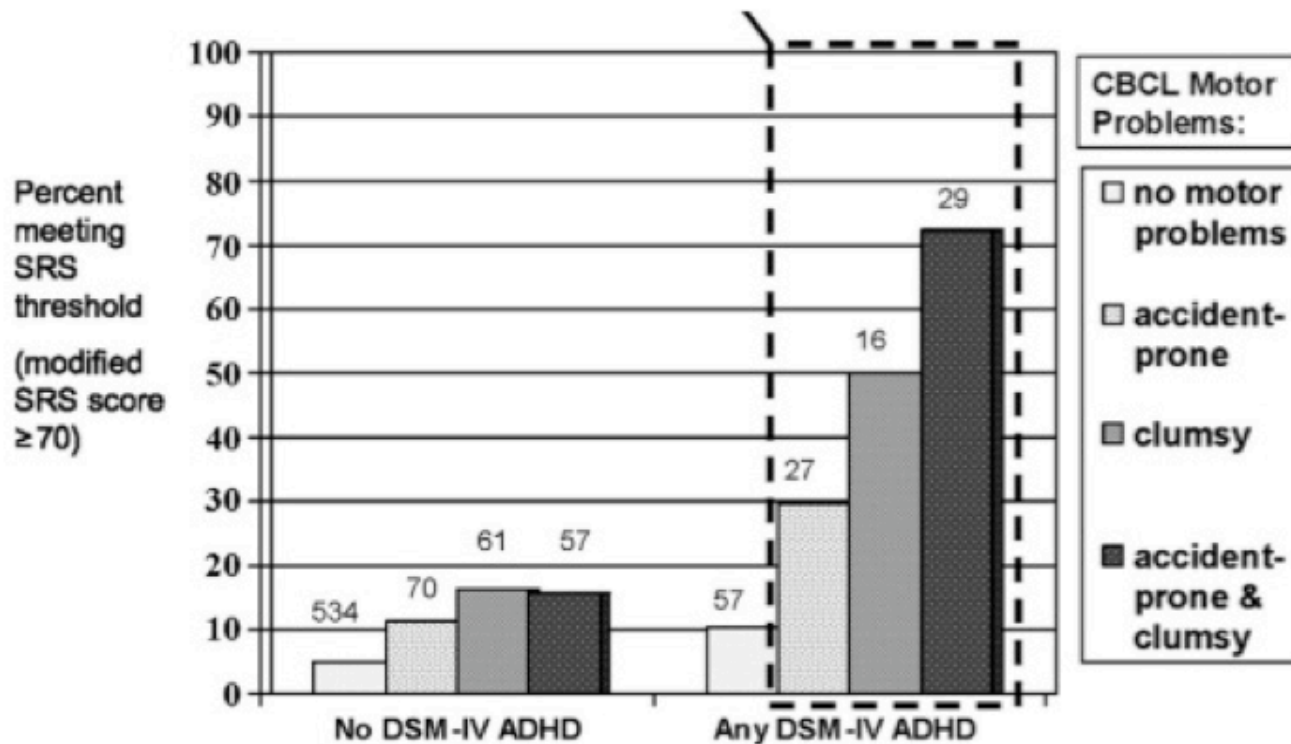
Symptoms or associated features common to several behavioral/ developmental disorders

- Hyperactivity
- Distractibility
- Irritability
- Impulsivity
- Social discomfort, isolation
- Disruptive behavior
- Repetitive behaviors/acts
- Sleep disturbance

- Moreover, multiple diagnoses often coincide:
- Tourette's with ADHD and OCD.
- ADHD and language delay
- ADHD and Anxiety

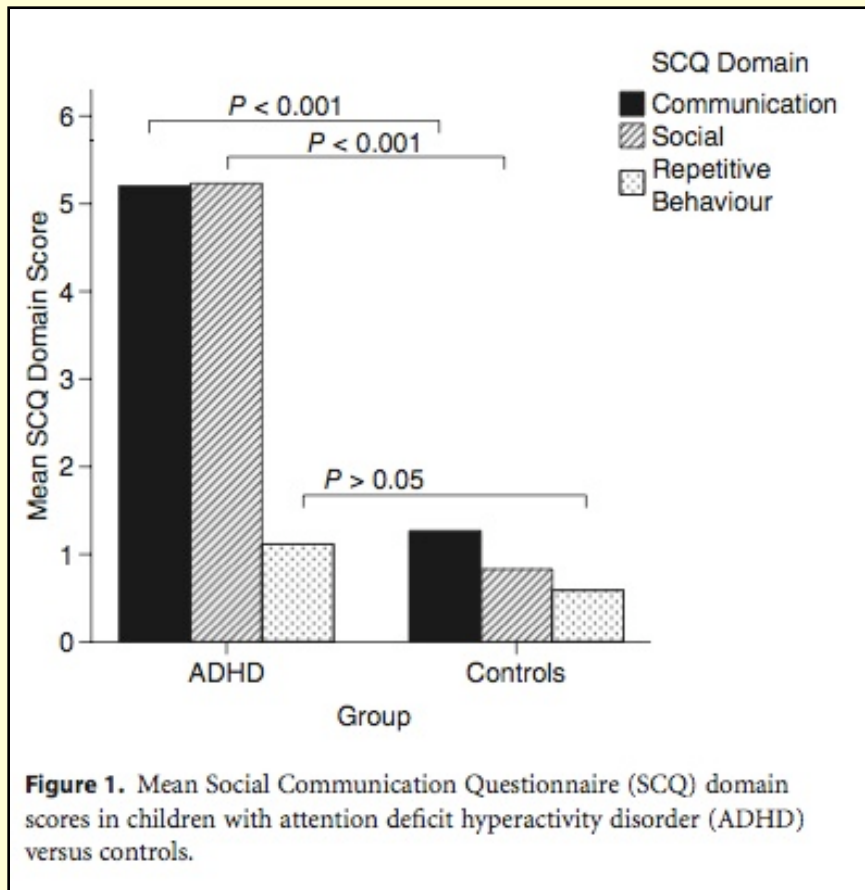
Clarifying ASD and other behavioral disorders

- Social Responsiveness Scale (SRS)
- Reiersen, Constantino, and Todd 2008
- N=821, 15% ADHD



Diagnostic overlap

Kochhar et al., 30 ADHD, 30 controls



- Children with ADHD had more ASD traits than controls, independent of IQ
- None had ASD, but 28–62% showed ASD traits
- ADHD had social and communication difficulties on the ASD measure, but not repetitive behaviors
- Unclear whether the social communication difficulties in ADHD are qualitatively different from ASD

- Choi & Esler, UMN clinic data:
- Social Communication Questionnaire (SCQ) Sensitivity and Specificity

	ASD	AUT	NONAUT	Non-ASD
Total Number	49	30	19	18
Mean Age(Months)	95.4	92.8	99.5	91.9
Mean SCQ Total Score	16.8	18.3	16.1	15.8
Male/Female Ratio	35/14	23/7	12/7	10/8

Table 2: Descriptive statistics summary. Total evaluation count, mean age, mean SCQ total score, and male to female ratio of ASD, AUT, NONAUT, Non-ASD group.

	ASD vs. Non-ASD	AUT vs. Non-ASD	NONAUT vs. Non-ASD
SCQ cut-off 15			
Sensitivity	.60	.57	.65
Specificity	.47	.47	.47
SCQ 80% Sensitivity Criteria			
Specificity	.29	.41	.12
SCQ 80% Specificity Criteria			
Sensitivity	.23	.30	.18

Table 3: SCQ cutoff criteria group comparisons. Sensitivity and specificity of ASD, AUT, and NONAUT vs. Non-ASD. Cutoff scores were determined to achieve 80% sensitivity and 80% specificity.

DSM-IV-TR interview

(Hartley & Sikora, 2009)

- Criteria that discriminated ASD from ADHD:
- Social relatedness:
 - Nonverbal behavior
 - Failure to develop peer relationships
- Communication
 - Delay/lack of speech
 - Stereotyped/idiosyncratic/repetitive speech
 - Lack of make-believe/imaginative play
- Restricted/repetitive:
 - None!

DSM-IV-TR interview

- Criteria that discriminated ASD from Anxiety:
- Social relatedness:
 - Nonverbal behavior
 - Lack of seeking to share enjoyment
 - Lack of social/emotional reciprocity
- Communication
 - Delay/lack of speech
 - Impaired conversational ability
 - Stereotyped/idiosyncratic/repetitive speech
 - Lack of make-believe/imaginative play
- Restricted/repetitive:
 - None!

CASE EXAMPLE

Tyler

ASD in Early Childhood

Social Communication

Limited Language

- **Reciprocity:** no joint attention, directing smiles, responding to others' attempts to engage
- **Nonverbal communication:** not pointing, no gestures, flat facial expression, infrequent or avoidant eye contact, use of others as tool
- **Relationships:** not watching other children; not seeking parents as a secure base; initiation of affection

Verbal/cognitive Skills Intact

- **Reciprocity:** inconsistent joint attention (e.g., strong interests only), consistent initiation of requests but not comments
- **Nonverbal communication:** limited pointing, gestures, facial expressions limited in range, limited or unusual eye contact
- **Relationships:** may watch but not seek out children; respond if another child has something interesting; expression of affection

ASD in Early Childhood

Repetitive Behaviors/Interests

Limited Language

- Any and all may be present
- Fixated interests may be less clear
- Child may prefer RRB to other activities
- Distress at interruption

Verbal/Cognitive Skills Intact:

- Any and all may be present (incl. fixated interests)
- Engages in RRB more in less structured activities
- May be easier to redirect

Background Information

- 3-year 0-month old boy
- Referred by pediatrician
- Early Childhood Special Education services under the primary eligibility category of ASD and secondary eligibility category under Speech or Language Impairment
 - 2 days per week for 3 hours
 - Receives speech and language and occupational therapy services
- Family questioning if a clinical diagnosis is also appropriate

Current Concerns

- Caregivers
 - Limited communication
 - Repetitive behaviors (e.g., spinning objects)
 - Activity level and limited safety skills
- Teacher
 - Limited communication
 - Difficulty learning new activities that are adult-directed
 - Difficulty following classroom rules
 - Limited interactions with peers

Tests Administered

- Mullen Scales of Early Learning
 - Visual Reception: impaired
 - Fine Motor: borderline
- Preschool Language Scale–5th Edition
 - Total Language: moderately–impaired
- Vineland Adaptive Behavior Scales–2nd Edition
 - Communication: moderately–impaired
 - Adaptive Behavior Composite, Daily Living Skills, and Socialization: mildly–impaired
 - Motor Skills: low average
- Toddler Autism Diagnostic Interview–Revised
- Autism Diagnostic Observation Schedule, Module 1

ADI-R

- First concerns at 18 months of age: language delays and repetitive behaviors/interests
- Social communication
 - Current says 3 single words/approximations; not purposeful
 - Mostly requests by guiding parents, giving items, and using their hand as a tool
 - Limited nonverbal communication
 - Occasionally shares enjoyment with his parents and sibling during highly preferred activities
 - Limited play skills
 - Not showing interest in peers; engages in some gross motor play with his sibling

ADI-R

- Restricted, repetitive patterns of behavior, interests, or activities
 - Turns on and off lights, overly focused on writing utensils, spins objects, and plays with parts of objects (i.e., car wheels)
 - Hand mannerisms and complex body movements
- Additional behaviors
 - Very active and difficulty remaining seated
 - Frequently attempts to run from caregivers
 - Disruptive and risk-taking behaviors

ADOS: Module 1

Observations:

- Social communication
 - Typically communicated using physical means
 - Giving objects or using someone's hand as a tool
 - Used 1 gesture, no pointing, no words
 - Unusual vocalizations
 - Infrequently made eye contact
 - Shared enjoyment during 2 activities
 - Directed a few smiles and an excited face
 - No instances of showing items, no initiation of joint attention and no response to joint attention
 - Limited engagement in play, no imitation

ADOS: Module 1

Observations:

- Restricted, repetitive patterns of behavior, interests, or activities
 - Visually inspected lights on toys
 - Spun objects, was preoccupied with parts of objects, and turned lights on and off
 - Strong interest in examiner's pen
 - Overly focused on some toys and tantrummed when they were removed
 - Jumped and flapped his arms
- Additional behaviors
 - Very active and climbed on furniture
- Score fell within the Autism range

Diagnoses

- Autism Spectrum Disorder
- Mixed Receptive–Expressive Language Disorder
- Continue to monitor for possible Intellectual Disability

General Recommendations for a 3 year old, first diagnosed with ASD

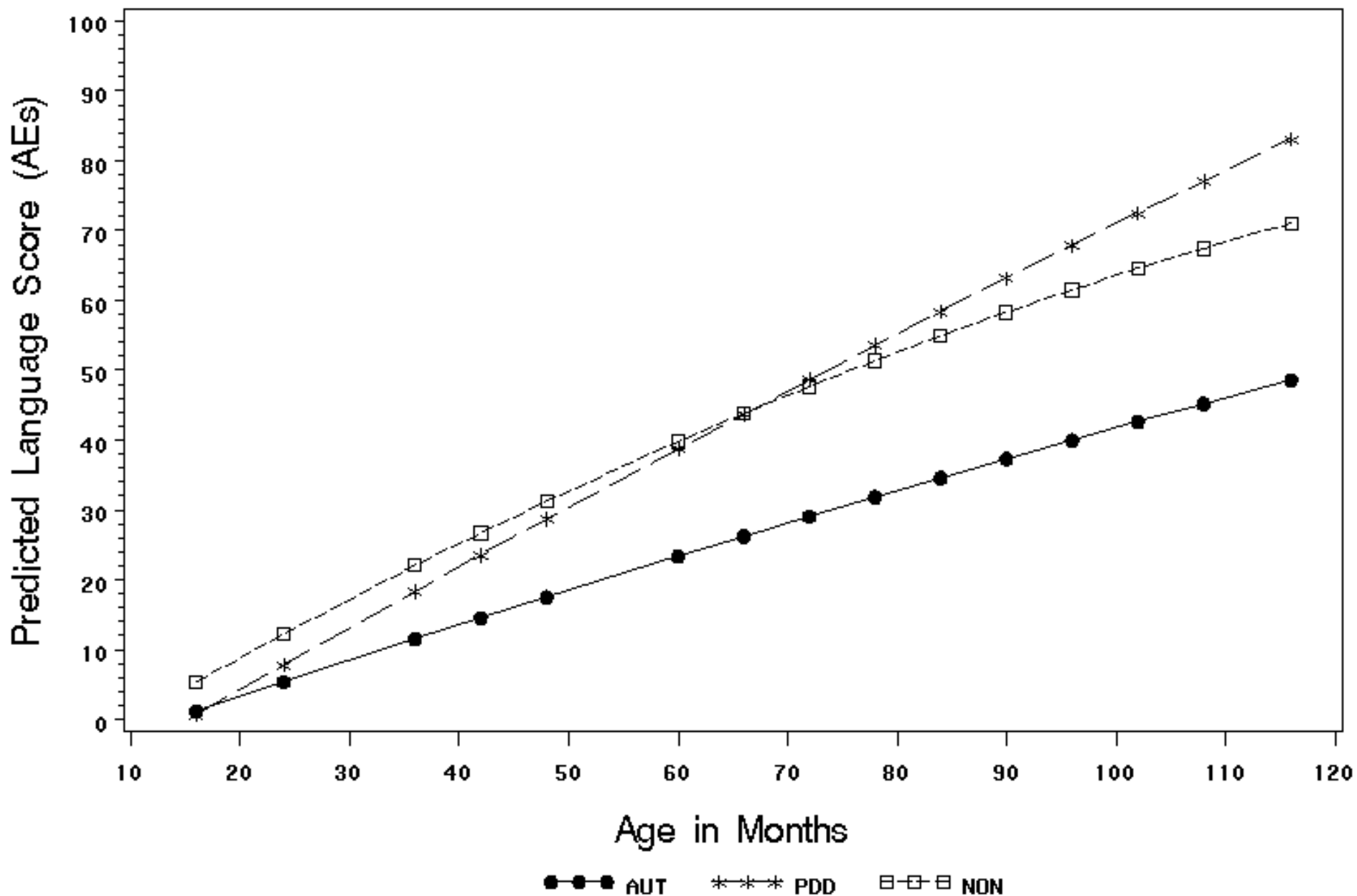
- Full-time intensive behavior intervention (e.g., ABA)
- Private speech-language and occupational therapy services
- Community resources (i.e., county social worker, financial planning)
- Genetic testing
- Re-evaluation annually until elementary school
- As indicated:
 - Medical follow-up
 - Complementary/alternative treatments

Prognosis

- Very much affected by language level and whether or not there is intellectual disability
- Usually a lifelong disorder, but many more students now attending regular school
- Many more young adults now employed at least part-time and living in the community

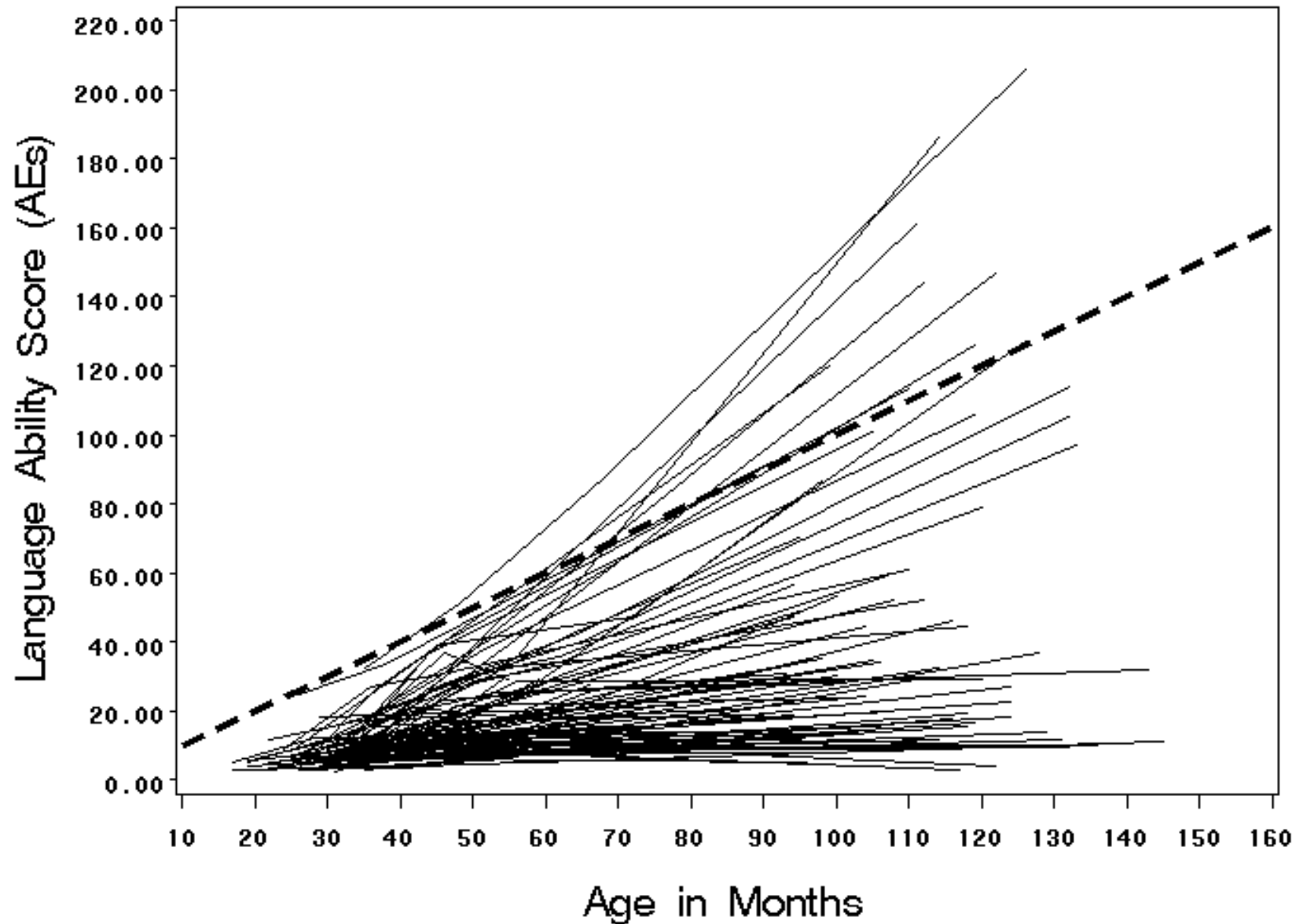
Predicted Language Trajectories by Age 2 Diagnosis

(with covariates included)



Individual Language Trajectories through Age 9 +

Autistic: Based on Age 2 Diagnosis



Summary



- ASD is a behavioral diagnosis, reliant on the skills and experience of the examiner
- Autism can be reliably diagnosed at 2 years by trained, competent specialists.
- Differential diagnosis is most difficult at the extremes of cognitive skills, behavior problems
- Questionnaires are widely used but of limited utility



Minnesota LEND

University of Minnesota Autism Spectrum Disorder Program

<http://lend.umn.edu/>

