# Autism Spectrum Disorder: Diagnosis, Assessment and Best Practices

Robin K. Rumsey, Ph.D.
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## Overview

- DSM-IV criteria
- Proposed DSM-V criteria
- Early identification
- Assessment of ASD
- Epidemiology/ Cultural Factors

## Facts About Autism

- It is estimated that 1 in 110 children born in the United States will be diagnosed with an autism spectrum disorder (1 in 70 boys).
- The cost per year of autism is estimated at 90 billion dollars (90% of the costs being in adult services).
- The cost of lifelong care can be reduced by 2/3 with early diagnosis and intervention.

## Facts About Autism

- 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.

## Variability of ASD

<table>
<thead>
<tr>
<th>Cognitive Skills</th>
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<tbody>
<tr>
<td>Severe = Gifted</td>
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<table>
<thead>
<tr>
<th>Social Interaction</th>
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<tr>
<td>Aloof = Passive = Active but odd</td>
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<table>
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<th>Communication</th>
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<tr>
<td>Nonverbal = Verbal</td>
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<table>
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<th>Behaviors</th>
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<tr>
<td>Intense = Mild</td>
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<table>
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<tr>
<th>Sensory</th>
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<tr>
<td>Hyposensitive = Hypersensitive</td>
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<tr>
<th>Motor</th>
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<td>Uncoordinated = Coordinated</td>
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Social behavior is not all-or-nothing

- **Social behaviors are not completely absent** (despite DSM wording)
- **Children with autism do show social behaviors** (e.g., eye contact, imitation, attachment)
- **Social behaviors occur less consistently across people and settings** (e.g., at different times, requiring greater effort)

Diagnostic Criteria for Autism

- **Social Interaction** — at least 2 of the following:
  - Marked impairment in the use of multiple nonverbal behaviors
  - Failure to develop peer relationships appropriate to developmental level
  - Lack of spontaneous seeking to share enjoyment, interests or achievements with others
  - Lack of social or emotional reciprocity

Diagnostic Criteria for Autism

- **Communication** — at least one of the following:
  - Delay in the development of spoken language
  - For those with adequate speech, marked impairment in the ability to sustain a conversation with others
  - Stereotyped and repetitive use of language or idiosyncratic language
  - Lack of pretend play

Diagnostic Criteria for Autism

- **Restricted interests/ repetitive behaviors** — at least one of the following:
  - Encompassing preoccupation
  - Inflexible adherence to nonfunctional routines or rituals
  - Stereotyped and repetitive motor mannerisms
  - Persistent preoccupation with parts of objects
Autistic Spectrum Diagnoses

- Autistic Disorder
- Asperger Syndrome
- Rett’s Syndrome
- Childhood Disintegrative Disorder
- Pervasive Developmental Disorder Not Otherwise Specified (PDD NOS)

Diagnosis

Qualifiers

- Onset in at least 1 domain before age 3
- Not better accounted for by other diagnosis

Draft Proposals for DSM V

- 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)

Draft Proposals for DSM V

- Three existing domains (socialization, communication, restricted/repetitive) will become 2 domains:
  - Social Communication
  - Restricted/repetitive behaviors (RRB’s)

Draft Proposals for DSM V

- For social communication domain, an individual must meet criteria for ALL of the following:
  - Deficits in social-emotional reciprocity
  - Deficits in nonverbal communicative behaviors used for social interaction
  - Deficits in developing and maintaining relationships, appropriate to developmental level

Draft Proposals for DSM V

- All individuals have or have had restricted interests and repetitive behaviors (at least 2 of 4)
  - Stereotyped or repetitive speech, motor movements or use of objects
  - Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior or excessive resistance to change
  - Highly restricted, fixated interests that are abnormal in intensity or focus
  - Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment
Draft Proposals for DSM V

- 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

DSM V Severity Levels

- **Level 3** (Requiring very substantial support)
  - Deficits in verbal and nonverbal social communication skills cause severe impairments in functioning, very limited initiation of social interactions, and minimal response to social overtures from others.

- **Level 2** (Requiring substantial support)
  - Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place; limited initiation of social interactions and minimal response to social overtures from others.

- **Level 1** (Requiring support)
  - Without supports in place, deficits in social communication cause noticeable impairments. Has difficulty initiating social interactions and minimal response to social overtures from others.

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).

Challenges of Diagnosing Autism Under 3 Years

- Increased behavioral variability at young ages.
- Overlapping symptoms with other developmental disorders (e.g., developmental delay, language delay/disorder).
- Limited applicability of current diagnostic systems and measures.

Early Identification

**Why?**

- Self-imposed social deprivation compromises behavioral and brain development
- Outcome evidence supports early intervention
  - Early identification $\rightarrow$ intense structured social input $\rightarrow$ More typical development

Models of Developmental Outcomes

**How early is “early” diagnosis?**

**1990’s**
*Can we diagnose autism in 2 year-olds?*

**2000’s**
*Can we diagnose autism in infants and toddlers?*
Diagnostic Disconnect

- The average age of parental concern is 17 months
  
  YET...

- The average age of diagnosis is 3 to 4 years

Retrospective Studies

- About 50% of parents recall having concerns about their child later diagnosed with ASD in the first year.

- Review of 1st birthday videos showed response to name and eye contact as distinguishing ASD from controls.

Prosperspective Studies

- Can follow children at higher risk for ASD to study early manifestations, both behavioral and neurobiological.

- E.g. Zwaigenbaum et al

Infant Sibling Study

- Autism Observation Scale for Infants (AOSI): Bryson, S., Zwaigenbaum, L., McDermott, C., Rombough, V. & Brian, J.

6 Months and Younger

- At 6 months no signs evident that predict later diagnosis

- Motor control differentiates controls from ASD in a small proportion of infants

- Strikingly, most infants later diagnosed with ASD look socially engaged at this age; indistinguishable from controls

12-14 Months

- AOSI scores in the risk range were a good predictor of ASD at 3 years in infants at high risk for ASD (15/17 children with elevated scores received an eventual diagnosis on the spectrum)

- Negative temperament and affect

- Delays in expressive and receptive language

- Subset of children experienced a decrease in IQ between 12-36 months (males; more severe)
Risk Markers on AOSI at 12 months
- eye contact
- visual tracking
- visual disengagement
- orient to name
- imitation
- social smile
- reactivity
- social interest and shared affect
- sensory-oriented behaviors

Red Flags of Autism Spectrum Disorder and Developmental Delays in the Second Year of Life
- Lack of showing
- Lack of coordination of nonverbal communication
- Lack of sharing interest or enjoyment
- Repetitive movements with objects
- Lack of appropriate gaze
- Lack of response to name
- Lack of warm, joyful expressions
- Unusual prosody
- Repetitive movements or posturing of body
- Lack of pointing
- Lack of playing with a variety of toys
- Lack of response to contextual cues
- Lack of communicative vocalizations with consonants

Project Overview - Nelson et al
- Longitudinal investigation
  - (3), 6, 9, 12, 18, 24 and 36 months (outcomes)
- Behavioral Measures
  - Mullen; Language; AOSI; temperament
- Growth Measures
  - Head Circumference; height/weight
- Experimental eye-tracking (faces; language)
- Electrophysiology (faces; speech; baseline EEG)

Visual Event-Related Potentials (ERP)
Design:
- Present alternating pictures of familiar face (mother) and unfamiliar face (stranger)
Summary: Visual ERP

- LRC infants show a developmental progression in their neural response to familiar and unfamiliar faces
  - No diff at 6 mos
  - Larger to mom at 9 mos
  - Larger to stranger at 12 mos

- HRA infants show no developmental transitions in their neural response to familiar and unfamiliar faces

ERP Response to Speech in Infants at Risk for Autism Spectrum Disorder

Lateralization of Language

- Typical infants show hemispheric lateralization by 9 months
- High risk infants fail to develop this by 12 months

Assessment for ASD

Same results found when participants with preliminary ASD diagnoses were removed

Full Evaluation Versus Screening

Screening
- Refer for diagnostic evaluation
- Clinical sample versus population sample

Complete evaluation
- Maximize accuracy of diagnostic classification
- Assess developmental levels
- Recommendations for treatment

Diagnosing ASD

- There is a difference between a medical diagnosis and educational identification of ASD!
- Educational ASD does NOT always = Medical ASD (and visa versa)
Dual Classification Considerations

- Can be very confusing & frustrating for parents
  - Terminology differences
  - Eligibility Criteria differences
  - Medical/Clinical vs. Educational Process
  - Timelines
  - Services available from each entity

Assessment for ASD

- Should include direct assessment of cognitive skills/development, language, adaptive functioning, and behavior whenever possible.
- Diagnosis should be based on parent interview (including developmental history), direct observation (Autism Diagnostic Observation Schedule) and, if possible, teacher observations.

Measures often used in diagnostic evaluations for ASD

**Autism 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)

**Other measures:**
- Adaptive: Vineland-II
- Cognitive:
  - Differential Abilities Scales (DAS-2)
  - Mullen Scales of Early Learning
- 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

Medical Work-Up

- Genetic Testing
  - High resolution chromosomes
  - Reflex to CGH
  - Fragile X
- Consider:
  - EEG
  - MRI
  - Lead
  - Vitamin D

Medical Issues

- Sleep problems 10-75%
- Gastrointestinal
  - 46%-86% vs. 17-24% general population
- Nutritional/Feeding
  - Selective diet/oral aversion, specific diets
- Seizures 25%
- Tics 9%
- ADHD 30-75%
- Affective disorders
  - Anxiety, depression
- Challenging behaviors
  - Risperidone (irritability)

Levels of Training/Expertise

- Medical Diagnosis:
  - Licensed Psychologist (Masters or Ph.D.)
  - Physician (MD)
- Experienced clinician
ADOS Administration

• To be used by professionals who are very familiar with Autism Spectrum Disorders (ASD).
• The ADOS has been given by clinical psychologists, school psychologists, speech/language pathologists, psychiatrists, pediatricians, social workers, graduate students, and other Autism specialists.

Obtaining ADOS Competence

• Have prior education, training and experience that include extensive exposure to Autism and PDD.
• Take an in-person clinical training workshop, either through UMACC or WPS, or use the WPS training video and accompanying materials.
• Practice using the ADOS on cases that are not part of formal evaluations and bring yourself to complete familiarity with the assessment activities and complete confidence that you can apply the coding categories accurately.
• If a user is to be involved in formal research directed at producing articles for publication in peer-review journals, he or she must additionally take a research training workshop followed by exercises that establish item coding accuracy to a specific criterion.

Demographics and Cultural Considerations

• 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

Demographics of ASD

Autism Statistics

• Estimates of 2-6+ in 1,000 children for spectrum (was 4-5 in 10,000 for autism) (APA, 2000)
  – 1 in 250 (Yeargin-Alsopp, 2003)
  – 1 in 150 (CDC, 2007)
  – 1 in 91 (CDC, parent report study, 2009)
• Factors that complicate interpretation
  – changes in diagnostic practice
  – Increased awareness of the disorder
  – earlier diagnosis
  – educational identification

Evidence for cultural differences?

Prevalence studies

- US
- UK
- Denmark
- Iceland
- Korea

Cases per 10,000
Methodological differences account for differences in prevalence across countries

- Accuracy affected by increased child mortality, poverty, health
- Some cultures may group ASD under another category
- Inconsistent medical record-keeping
- Integration of children with disabilities into society can affect ascertainment

Differences by ethnicity: US

- Medicaid-eligible children in Philadelphia:
  - White children diagnosed 1.5 years earlier than African American and 2.5 earlier than Latino
  - African Americans spent more time in treatment before diagnosis
  - African Americans 3x more likely to be misdiagnosed with a behavioral disorder before being accurately diagnosed with ASD
- Parent reporting?
- Prejudices and misconceptions among clinicians?

Interpretation of Behavior

- Medicaid-eligible children in Philadelphia:
  - White children diagnosed 1.5 years earlier than African American and 2.5 earlier than Latino
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Translating social symptoms across cultures

- Little is known
- India:
  - Language delays aren’t considered a core feature, because boys talk later than girls
  - Social symptoms recognized earlier due to emphasis on social conformity
- Saudi Arabia:
  - First concern is usually language delays
  - Girls diagnosed later due to societal expectations that boys are more outgoing
- Hmong:
  - Anecdotally, may be diagnosed later due to belief that children should be free to develop at their own course in early childhood

Cultural Perspectives

- Individualism v. collectivism
- Beliefs about causes

“People who see our children say, “Why is a normal child acting so strangely? Aren’t the parents sending him to school?” Children with autism often misbehave, so the first thing you think of when you see a kid with bad discipline is that there is a problem with their school or ability to learn or that they have a bad mother. I really think the parents of children who are not severely physically-disabled, like the parents the children with autism I know, suffer more than the parents of physically-handicapped children.”

--Mother from Seoul, quoted in R. Richard Grinker, Unstrange Minds: Remapping the World of Autism.

Case examples of Indian families’ description of their child’s symptoms

- A parent with a 3-year-old girl who did not relate socially with peers her age considered her a mature child because she responded well to adults.
- A mother was not alarmed that her son was still not speaking at 4 years because Indian boys talk later
- A child who keeps quiet and to oneself was perceived as a good child because he or she is trouble-free.
Genetic factors may contribute to variations across cultures
- ASD may have different liability alleles for different populations
- What is learned from one population may not transfer to another
- Consanguinity
- Environment and risk factors interact with genetics

Immigration as a risk factor?
- London study:
  - Caribbean and African immigrants 8-10x higher rate
  - Asian 2-4x higher
- Sweden:
  - Increased prevalence of ASD among Somali immigrants
  - All had co-occurring Intellectual Disability
- Stress of immigration, or related to factors within the home country?

Characterization of Somalis in MN
- Higher proportion of Somalis received autism preschool services in Minneapolis than other ethnic groups
- Anecdotally characterized as having more ID

The translation process for diagnostic tools (ADOS)
- Translation and back-translation
- Appropriateness of tasks and materials

Take-home tips
- Evaluate fluency in English as well as native language
  - Standardized measures may not apply
- Nonverbal testing—adapted for ASD
- Can give the ADOS, but not score it
- Rely on parent descriptions of their experiences, their child’s skills
  - Clinician’s job is to evaluate whether the behaviors described fit the pattern
- Trained, experienced interpreters

The End
rumse002@umn.edu