



# Autism Spectrum Disorder: History and Diagnostic Considerations

Sarah E. Lea, PhD

## Learning Objectives

- Describe the historical context of the DSM-5TR diagnostic criteria for ASD
- Discuss and identify current diagnostic criteria for ASD.
- Explain common age-related considerations in ASD assessment
- Identify and describe common comorbid diagnoses associated with ASD

## A Brief Historical Overview

- In 1867, Henry Maudsley described “insanity in children” in his text devoted to physiology and “pathology of the mind.”
  - Some of these descriptions appeared consistent with the current diagnostic criteria of ASD
- The German word “autismus” was coined by Swiss psychiatrist Eugen Bleuler
  - The word is derived from Greek “autos” (self) and “ismos” (suffix of action or of state)
  - Bleuler first used the term to describe “idiosyncratic, self-centered thinking that led to withdrawal into a private fantasy world”

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## A Brief Historical Overview

- In 1943 Leo Kanner introduced the modern concept of “autism” in a article published in the journal *Nervous Child*
  - Suggested children with autism “live in their own world, cut off from normal social intercourse”
  - Noted additional features such as “problems with symbolization, abstraction, and understanding meaning.” Also described these children as ridged, inflexible, and reacting negatively to changes in routine or in the environment
  - Kanner was the first to hypothesize that autism is distinct from schizophrenia, “representing a failure in development, not a regression.” He was also the first to suggest that autism is a genetically driven condition
    - Prior to this, psychoanalytic theory was suggesting autism was caused by inappropriate parenting

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## A Brief Historical Overview

- In 1944 physician Hans Asperger described a syndrome he called “autistic psychopathy”
  - Claimed he was unaware of Kanner’s work ☺
  - Syndrome would come to be known as Asperger’s disorder
- In 1908 Theodore Heller, a special educator, described a condition in which children displayed typical development for a number of years and then suffered a profound regression in functioning and development
  - Became known as Heller’s syndrome and Childhood Disintegrative Disorder in the DSM-IV-TR
- In 1966 pediatrician, Andreas Rhett first observed females with a developmental disorder characterized by a short period of normal development followed by a multifaceted form of intellectual and motor deterioration
  - Became known as Rhett’s Disorder in DSM-IV-TR

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## A Brief Historical Overview-DSM

- Until the 1970s autism was considered a form of schizophrenia with “childhood schizophrenia” the only term available to describe children with autism in both the 1<sup>st</sup> and 2<sup>nd</sup> editions of the DSM
- Cantwell, Baker, & Rutter (1980); DeMyer, Hingtgen, & Jackson (1981); and the convergence of the DSM and ICD classification systems led to the development of autism as it’s own diagnostic classification
- DSM-III (1980) first introduced “infantile autism”
  - However, specific symptoms and criteria were not defined
- DSM-III-R included more detailed and concrete descriptions of specific behaviors. Further the “lifelong” nature of the diagnosis was recognized
  - Change from “infantile autism” to “autistic disorder”
- DMS-IV Asperger’s disorder was included

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## DSM-5

- It had been anticipated in the DSM-IV the presence or absence of a history of language delay would be the differentiating factor between a diagnosis of autistic disorder and Asperger's disorder
  - It was originally assumed children with early language delays would differ in outcomes/presentation
  - However, what was considered "language delay" was not agreed upon
  - When language delay was classified as not meeting early developmental milestones (as determined by parent report) neither differences in outcome or presentation was present when controlling for current language functioning
  - Problems also emerged in that individuals were automatically given a diagnosis of Asperger's if they were perceived as having "milder" symptoms regardless of if they had a language delay or not.
  - Children who otherwise met criteria for ASD, but had strength in language skills were given "NOS" diagnoses
  - PDD- NOS had started becoming a "catch all" for children who had social challenges both with and without autism symptomology

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## DSM-5: The changes

- ASD now includes all individuals who received any former PDD diagnosis of autistic disorder, Asperger's, childhood degenerative disorder, or PDD-NOS
  - Rhett's syndrome is now diagnosed with genetic testing and is removed from DSM
  - Genetic or neurological disorders can be added as specifiers to ASD diagnosis
- DSM-5 collapsed Social Impairment and Communication Impairment domains in the DSM-IV-TR into 1 domain
- Language impairment is no longer a part of the diagnostic criteria as evidence suggests that language delay is not a unique or universal feature of ASD
  - Language level is still a significant factor in ASD and should be assessed

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## DSM-5TR: The changes

- Under criterion A, which describes differences in social communication and social interaction, the phrase “as manifested by the following” has been revised to read “as manifested by all of the following.”
- In addition to these social communication and interaction differences, at least two of four types of restricted or repetitive behaviors must also be present to meet the DSM-5-TR diagnostic criteria.
- The DSM-5-TR asks whether the autism diagnosis is associated with “another neurodevelopmental, mental, or behavioral problem.” Broadening this specifier allows clinicians to include information about associated problems that affect well-being but that may not be classified as disorders.

## DSM-5TR Criteria

- Deficits in Social-Emotional Reciprocity
  - Little to no initiation of social interaction, no sharing of emotions, reduced or absent imitation of others, one-sided language lacking reciprocity and used to request or label rather than comment, share feelings or converse, difficulties processing and responding to complex social cues
    - Struggle is apparent in novel or unsupported situations. May feel anxiety and displace considerable effort in doing what is “socially intuitive” for others
    - Social interactions might be labeled as exhausting or impact concentration

## DSM-5TR Criteria

- Deficits in Nonverbal Communicative Behaviors
  - absent/reduced/atypical eye contact/gestures/facial expressions/body orientation/speech intonation
  - Impaired joint attention-lack of pointing, showing or bringing objects, sharing interest with others, failure to follow point or gaze
  - Lack of expressive gestures or odd, wooden, or exaggerated body language

## DSM-5TR Criteria

- Deficits in, maintaining, and understanding relationships
  - Should only be judged based on norms for age, gender, and culture
  - Absent/reduced/atypical social interest, rejection of others, passivity, overt aggression, lack of shared social play/imagination, fixed rules in play, struggle to understand context of social behavior, understanding irony or "white lies"
  - May prefer solitary activities or preference for younger or older peers
  - Possible desire for relationships without complete or realistic idea of what it entails

## DSM-5TR Criteria

- Stereotyped or repetitive motor movements, use of objects, or speech
  - Hand flapping, finger flicking, spinning objects, lining up objects, echolalia, use of "you" when referring to self, stereotyped use of words, phrases, or prosodic patterns
- Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behaviors
  - Resistance to change (even small changes), ridged thinking, pacing, repetitive questioning
- Highly restricted, fixated interests that are abnormal in intensity or focus
  - Spending hours on interest, odd interest, etc.
- Hyper or hypo reactivity to sensory input or unusual sensory aspects of the environment
  - Extreme response to specific sounds/textures, excessive smelling/touching, fascination with lights/sounds, indifference to pain/heat/cold, food restrictions

[https://www.youtube.com/watch?v=E-XgK\\_LaFKI](https://www.youtube.com/watch?v=E-XgK_LaFKI)

<https://www.youtube.com/watch?v=ZpoYOgjJT8>

## Social (pragmatic) Communication Disorder

- New diagnosis in the DSM-5 that is not considered ASD
- Was developed out of concerns that some previously diagnosed with Asperger's or PDD-NOS might no longer meet criteria for revised ASD criteria
- Data is currently mixed and ASD must be thoroughly ruled out before diagnosing SCD
- Should not be used as a "catch all" category that PDD-NOS was previously used for

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

Feedback has been mixed on  
DSM-5 and DSM-5TR changes.  
What are your thoughts?



## Age Related Issues in ASD assessment

- Specific focus and tools for ASD assessment vary significantly depending on age of person being assessed.
  - Literature notes 5 developmental stages in assessment of ASD
    - Early Childhood (up to age 3)
    - Preschool (3-5 years)
    - Elementary School (6-11 years)
    - Middle/High School (12-17)
    - Adulthood (18+)
- Typically, assessment of young children focuses on diagnosis and assessment at later ages focuses on measuring skills

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Early Childhood/Preschool

- Although assessment tool exist to reliably diagnose ASD by age 2, the average age of diagnosis is about age 4
- The American Academy of Pediatrics (AAP) recommends all children be screened for ASD at 18, 24, and 30 months using standardized measures
- Accurate diagnostic terms should be used and explained and questions about the child's future developmental course should be answered as fully as possible.
- Information about educational and treatment options should be part of the initial diagnostic assessment
- Being aware of parent readiness/ ability to hold and process recommendations and next steps

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Early Childhood/Preschool: diagnostic tools

- First Year
  - Behaviors in first year that are strongly associated with ASD
    - Decreased social responsiveness (responding to name, looking at people, joint attention)
    - atypical sensory-regulatory behaviors (increased mouthing objects, unusual visual attention patterns, increased irritability)
  - Autism Observation Scale for Infants
    - Semi structured play based measure
  - First Year Inventory
    - Parent report

## Early Childhood/Preschool: diagnostic tools

- Second Year
  - Autism Diagnostic Observation Schedule (ADOS-2)
    - “gold standard” direct observation measure
  - Modified Checklist for Autism in Toddlers (M-CHAT)
  - Screening Tool for Autism in Two-Year-Olds (STAT)
    - Play-based observation
  - Bayley Scales of Infant and Toddler Development (Bayley-III)
    - “gold standard” for assessment of developmental delays in 12-24 months

## Early Childhood/Preschool: diagnostic tools

- Ages 2-3
  - Childhood Autism Rating Scale (CARS-2)
    - Less expensive/ complicated than ADOS
  - Autism Diagnostic Interview- Revised (ADI-R)
    - Administration time of 2 hours
- Age 4+
  - Social Communication Questionnaire (SCQ)
    - Parent report measure using questions for ADI-R that were found to be most associated with ASD
  - The Vineland Adaptive Behavior Scale, Second Edition

## Elementary School Age

- Often at this age assessment (or reassessments) focus on confirming ASD diagnosis and/or determine functioning abilities/impairment
- Reassessment occurs to support/modify/evaluate a wide range of interventions
- Functional skills assessments in multiple settings are often beneficial
  - Social skills, routine evaluations, transitions, toilet training, etc
- Note: research suggests females with ASD without intellectual impairment often go undiagnosed at this age

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Elementary School Age: diagnostic tools

- Child Behavior Checklist (CBCL)
  - Broadband rating scale
- Social Responsiveness Scale (SRS-2)
  - measure social skills deficits ages 4-18

## Middle/High School Age

- Reassessments for may focus on questions such as “what new supports/instruction is needed?” “what supports may be needed in future?” “what services are they eligible for?”
- Could be required for college preparation or for publicly funded programs such as SSI, Medicaid/Medicare, and Voc Rehab
- Often children who are first diagnosed at this age have been involved in treatment of some sort for years, struggling with diagnostic confusion

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Adulthood

- Reassessment in adults tends to focus on identifying needed supports and qualification for additional public services
- Individuals whose first assessment for ASD occurs in adulthood tend to fall into two categories
  - Those with severe developmental delay
  - Those with high cognitive and adaptive functioning

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Age Related Issues in ASD assessment

- A recent meta-analysis (35 studies, covering 55 samples from 35 countries, n = 66,966 individuals with ASD) found a mean age at diagnosis of 60.48 months (~5 years) with a range of 30.90–234.57 months (2.5-19.5 years).
- A large number of adults samples were included in this meta-analysis
- Let's talk about age and ASD assessment
  - Many diagnostic centers have limitations in this area
  - What are the pros/cons of late adolescent or adults ASD assessment?
  - What are the practical implications?

van't Hof, M., Tisseur, C., van Berckeleer-Onnes, I., van Nieuwenhuyzen, A., Daniels, A. M., Deen, M., ... & Ester, W. A. (2021). Age at autism spectrum disorder diagnosis: A systematic review and meta-analysis from 2012 to

## Comorbid Diagnoses and Autism Spectrum Disorder

- The issues of comorbidity and ASD has become increasingly important as the DSM-5/TR no longer excludes most additional diagnoses in individuals with ASD
- Some studies have found that over 70% of children with ASD met criteria for another diagnosis and that up to 40% may have 2 or more comorbid conditions
- Children with ASD have been found to have more psychiatric comorbidity than children with ID alone
- Comorbidity should be considered when
  - Sign of symptoms not part of ASD diagnosis
  - Changes in functioning from baseline or when existing symptoms is increased
  - When previously effective interventions are no longer effective

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Comorbid Diagnoses and Autism Spectrum Disorder

- It should be noted that instances of “false” comorbidity are also possible
  - Referral bias
  - Misunderstanding of ASD diagnostic criteria
  - Diagnostic overlap
  - One diagnosis representing an early manifestation of another diagnosis

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Challenges in Assessing Comorbid Conditions

- Standardization and Norming Issues
  - Most behavior checklists and diagnostic interview tools were standardized on individuals without ASD and/or ID
- Insight and Self-Report Problems
  - Those with ASD may struggle to talk about emotions or internal experiences
  - Often self report is “gold standard” in mental health diagnoses
  - Response perseveration
  - Some data does suggest that those with “higher functioning” ASD have larger capacity for introspection and self report

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Challenges in Assessing Comorbid Conditions

- Differences in Symptoms Manifestation
  - Individuals with ASD may not demonstrate certain symptoms related to other mental health diagnoses
    - Changes in sleep or eating behaviors, ruminations, somatic complaints, emotion based symptoms
- Potential Solutions
  - Focus on changes in behavior from baseline
  - Appearance of new behaviors or loss of previously achieved skills/functioning

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## Differential Diagnosis VS Comorbidity

- Examine the developmental history for consistency of symptoms over time and pervasiveness across situations
- Social and communication limitations AND odd or repetitive behaviors expresses consistently throughout the lifetime
  - No other condition has the combination of the above

Goldstein, S., & Ozonoff, S. (Eds.). (2018). *Assessment of autism spectrum disorder*. Guilford Publications.

## ASD and Depression

- Studies indicate the rates of depression in children and adults with ASD to be between 19 and 56%
- National Survey of Children's Health found rates of comorbid well above the general population in both children (12.5%) and adolescents (31.3%) with ASD
- In individuals with ASD the presenting complaint is often not mood related , but rather new or worsening aggression, agitation, self-injury, increased compulsive behaviors, hypoactivity, sleep problems, loss of bowel control, loss of interest in special topic, and overall deterioration in everyday functioning across environments



## ASD and Bipolar Disorder

- Approximately 2% of adolescents in the general population meet criteria for bipolar I. The prevalence rate of bipolar I in ASD is unknown due to lack of studies.
- Reported rates of mania in children/teens with ASD ranges from 2-31% and 6-21% in adults
- Symptoms of bipolar disorder can be difficult to parse apart from ASD symptoms.
- Sleep impairment may be an indicator if detailed sleep history is able to be obtained.
- Key is to evaluate mood relative to baseline behaviors

## ASD and Anxiety

- A large NSCH epidemiological study of children with ASD found that 18.8% of preschoolers, 39.4% of school-aged children, and 48.9% of teens with ASD met criteria for an anxiety diagnosis.
  - Rates well above general population
- Anxiety seems to increase with age in individuals with ASD.
- Some studies suggest specific phobias are the most common co-occurring diagnoses
- Anxiety in ASD appears more “trait-like” than “state-like”
- Irritability is most common reported symptom of anxiety in ASD

## ASD and OCD/related disorders

- No prevalence studies of co-occurrence of ASD and OCD/related disorder have been conducted
- ASD symptoms can often be confused with symptoms of OCD
- Differentiating for ritualized behaviors and compulsions can be difficult
  - In ASD rituals often appear to bring pleasure and are less ego dystonic than in OCD
  - In ASD these behaviors are not conducted in order to counteract intrusive thoughts or reduce anxiety

## ASD and ADHD

- DSM-5 first to allow dual diagnosis of ADHD and any pervasive developmental disorder
- NSCH study of children with ASD reported 21.6% of preschoolers, 44.6% of school-aged children and 52.6% of teens also met criteria for ADHD
- Dual diagnoses are often given in clinical setting due to the level of impairment associated with ADHD-like symptoms and need for treatment
- “Overfocus” and internal distractibility may be more of a symptoms of ASD while “under focus” and external distractibility may be more associated with ADHD

## ASD and Tic Disorders

- Prevalence rate of Tourette's disorder is reported to be 0.77% and of transient tic disorder to be 3%. Rates are consistently higher in boys than girls and in children than adults.
- Recent studies indicate in clinic referred children with ASD 23% met criteria for chronic motor or vocal tic disorder and 18% met criteria for Tourette's. In clinic referred adults, 20% met criteria for chronic tic disorders
- Onset of tic disorder is typically age 6-7
- In tic disorders, behaviors are rapid, involuntary, nonrhythmic, sudden, interrupt flow of behavior/speech, and are preceded by an unpleasant sensation or urge
- In tic disorders behaviors tend to involve face, neck, arms, and shoulders. While in ASD behaviors tend to involve hands, fingers, and whole body

## ASD and Psychosis

- No epidemiological studies on schizophrenia spectrum disorders and ASD
- Recently there has been an increased interest in the overlap between the schizophrenia spectrum and autism spectrum
- Pragmatic language deficits in ASD may present like a thought disorder, lack of motivation in certain areas may be confused with anhedonia, sensory sensitivity and preoccupations may be confused for hallucinations
- The relationship between ASD and childhood onset of schizophrenia is not well understood as a high percentage of children with schizophrenia are first diagnosed with ASD
  - Prodromal schizophrenia has most overlap with ASD like presentation

