
AUTISM SPECTRUM DISORDERS

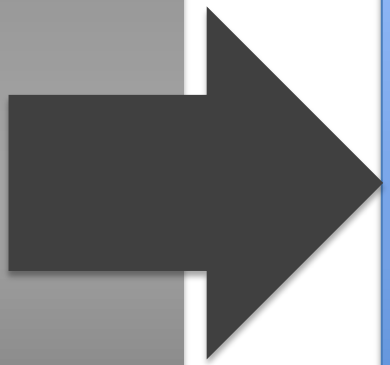
From *First Aid for the Psychiatry Clerkship*:

A 3-year-old boy is brought in by his parents because they think he is deaf. According to the parents, he shows no interest in them or anyone around him and only speaks when spoken to directly. He often takes his toys and lines them up in a straight line. His hearing tests are normal. *Think: Autism.*

DSM IV

PERVASIVE DEVELOPMENTAL DISORDERS

- AUTISTIC DISORDER
- RETT'S DISORDER
- CHILDHOOD DISINTEGRATIVE DISORDER
- ASPERGER'S DISORDER
- PDD NOS
- CHILDHOOD AUTISM
- KANNER'S AUTISM
- HIGH-FUNCTIONING AUTISM
- ATYPICAL AUTISM



DSM V

AUTISM SPECTRUM DISORDER

RECORDED WITH:

- ASSOCIATED KNOWN MEDICAL OR GENETIC CONDITION OR ENVIRONMENTAL FACTOR
- SEVERITY SPECIFIED
- INTELLECTUAL IMPAIRMENT SPECIFIED
- LANGUAGE IMPAIRMENT SPECIFIED

If diagnostic criteria not met, evaluate for SOCIAL COMMUNICATION DISORDER

DIAGNOSTIC CRITERIA

A. **Persistent deficits in social communication and social interaction across multiple contexts as manifested by the following, currently or by history**
   1. Deficits in social-emotional reciprocity
      - Failure of normal back-and-forth conversation
      - Reduced sharing of interests, emotions, or affect
      - Failure to initiate or respond to social interactions
   2. Deficits in nonverbal communication behaviors used for social interaction
      - Poorly integrated verbal and nonverbal communication
      - Abnormalities in eye contact and body language
      - Deficits in understanding or use of gestures
      - Ranging to a total lack of facial expressions and nonverbal communication
   3. Deficits in developing, maintaining, and understanding relationships
      - Difficulties adjusting behavior to suit various social contexts
      - Difficulties sharing imaginary play
      - Difficulties making friends
      - To absence of interest in peers

Table 2 Severity levels for autism spectrum disorder

Severity level	Social communication	Restricted, repetitive behaviors
Level 3 "Requiring very substantial support"	Severe 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. For example, a person with few words of intelligible speech who rarely initiates interaction and, when he or she does, makes unusual approaches to meet needs only and responds to only very direct social approaches.	Inflexibility of behavior, extreme difficulty coping with change, or other restricted/repetitive behaviors markedly interfere with functioning in all spheres. Great distress/difficulty changing focus or action.
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 reduced or abnormal responses to social overtures from others. For example, a person who speaks simple sentences, whose interaction is limited to narrow special interests, and who has markedly odd nonverbal communication.	Inflexibility of behavior, difficulty coping with change, or other restricted/repetitive behaviors appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts. Distress and/or difficulty changing focus or action.
Level 1 "Requiring support"	Without supports in place, deficits in social communication cause noticeable impairments. Difficulty initiating social interactions, and clear examples of atypical or unsuccessful responses to social overtures of others. May appear to have decreased interest in social interactions. For example, a person who is able to speak in full sentences and engages in communication but whose to-and-fro conversation with others fails, and whose attempts to make friends are odd and typically unsuccessful.	Inflexibility of behavior causes significant interference with functioning in one or more contexts. Difficulty switching between activities. Problems of organization and planning hamper independence.

B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history

1. Stereotyped or repetitive motor movements, use of objects, or speech

(e.g. simple motor sterotypies (hand flapping, finger flipping), lining up toys or flipping objects, echolalia, idiosyncratic phrases)

2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (e.g. extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat the same food every day)

3. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g. strong preoccupation to or preoccupation with unusual objects, excessively circumscribed or perseverative interests)

4. Hyper or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment (e.g. apparent indifference to pain/ temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement)

C. Symptoms must be present in early developmental period (but may not become fully manifest until social demands exceed limited capacities or may be masked by learned strategies later in life)

D. Symptoms cause clinically significant impairment in social, occupational, or other areas of current functioning

E. These disturbances are not better explained by intellectual disability or global developmental delay. Intellectual disability and autism spectrum disorder frequently co-occur; to make comorbid diagnoses of autism spectrum disorder and intellectual disability, social communication should be below the general developmental level.

Specify if:

- With or without accompanying intellectual impairment
- With (nonverbal/single words/phrase speech) or without accompanying language impairment
- Associated with a known medical (epilepsy), genetic condition (Rett syndrome, Fragile X, Down Syndrome) or environmental factor (maternal exposures)
- Associated with another neurodevelopmental, mental or behavioral disorder (ADHD; developmental coordination disorder; disruptive behavior, impulse control or conduct disorders; anxiety, depressive or bipolar disorders; Tourette's; self-injury, feeding, elimination, or sleep disorders)
- With catatonia

From *Blueprints Psychiatry*:

"In Asperger's disorder, there are **no** delays in language or cognitive, self-help, environmental curiosity or adaptive behavior domains."

Example question:

A 12 yo girl presents with her foster mother for a routine pediatric visit. The girl seems quiet, and she does not make eye contact with you as you enter the room. Her foster mother says that the girl has been having difficulties socially in school. She has trouble making friends and often has conflicts with classmates. She has trouble seeing the "big picture" of problems that face her, and she often gets lost in the details of projects that she is assigned. She is awkward in her interactions. Her foster mother says she can be quite verbally articulate and writes extremely well. You query her for depressive or psychotic symptoms and she denies either. Once you ask her about one of her projects, she tells you about the intricacies of building a radio from scratch. She engages with you briefly but still appears uncomfortable. She denies any significant anxiety. The most likely diagnosis is:

a. Asperger's syndrome

b. Autism

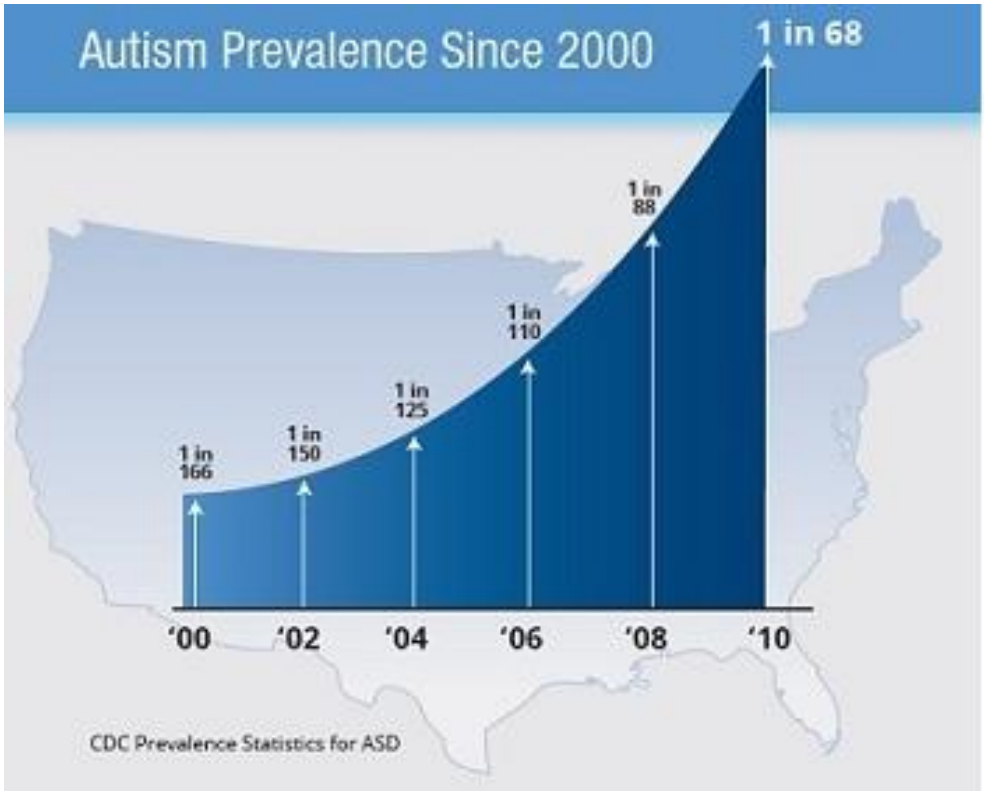
c. Avoidant personality disorder

d. Anxiety disorder NOS

e. PTSD

Prevalence

- M: F - 5:1
- White > Black=Asian/Pacific Islander > Hispanic
- 1980s – 0.5 per 1000
  1990s – 2 per 1000
  2014 – 1 per 68
- 31% had IQ<=70
  23% had IQ = 71-85
  46% IQ > 85
- Other associated conditions: Tuberous Sclerosis, Fragile X, 15 q abnormalities, Rett syndrome, Smith-Lemil-Opitz syndrome, Various metabolic conditions



From the CDC's *Community Report on Autism 2014*

Signs and Symptoms

A child with ASD might:

- ✓ Not respond to his or her name by 12 months of age (for example, appear not to hear).
- ✓ Not point at objects to show interest by 14 months of age (for example, point at an airplane flying over).
- ✓ Not play "pretend" games by 18 months of age (for example, pretend to "feed" a doll).
- ✓ Avoid eye contact and want to be alone.
- ✓ Have trouble understanding other people's feelings or talking about his or her own feelings.
- ✓ Have delayed speech and language skills (for example, use words much later than siblings or peers or not use words to communicate).
- ✓ Repeat words or phrases over and over.
- ✓ Give unrelated answers to questions.
- ✓ Get upset by minor changes in routine (for example, getting a new toothbrush).
- ✓ Have obsessive interests (for example, having a very strong interest in trains that is difficult to interrupt).
- ✓ Flap his or her hands, rock his or her body, or spin in circles.
- ✓ Have unusual ways of playing with or using objects, such as spinning or lining them up repeatedly.

Pathogenesis

- Not vaccines!
- Concordance among monozygotic twins: 36-96%; Siblings: 3-10%
- Multiple genes + environmental factors
  - Genetic abnormalities
  - Global and functional abnormalities in serotonin synthesis
  - Enlarged cerebral gray and white matter by 2.5 years particularly in the frontal/temporal/cingulate cortices
  - Maternal exposures: Maternal metabolic conditions, Valproate, Probably not SSRIs
  - Increased parental age (de novo mutations and/or alterations in genetic imprinting)

Treatment

- Chronic condition
- Treat to maximize functioning and improve the quality of life
- Behavioral/educational interventions targeting the core symptoms: Deficits in social communication and social interaction and Restricted patterns of behavior, interests, activities
  - Ex. TEACCH in NC, many others
- Medication to treat:
  - Hyperactivity, inattention, impulsivity: methylphenidate, guanficine, atomoxetine
  - Aggression, outbursts and self-injury: risperidone, aripiprazole
  - Anxiety: SSRI
  - Obsessive compulsive behaviors, repetitive behaviors: SSRIs, clomipramine, risperidone, valproate
  - Depression: SSRI/SNRI, not much trial data
  - Sleep dysfunction: sleep hygiene, behavioral intervetions
  - Only risperidone and aripiprazole are approved by the FDA

Prognoses

- For more able individuals with ASD
- Asperger (1944) – special skills or interests ‘eventually led social integration’
- Looking at six small sample studies from the 80s-90s
- College educated: 5-32%
- Living (semi-)independently: 16-50%
- In paid work: 5-55%
- “Good” outcome: 16-44%
- “Poor” outcome: 0-74%

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