

Autism Spectrum Disorder

DSM-5-TR 299.00 | ICD-10-CM F84.0

Persistent social communication deficits plus restricted, repetitive behaviors, present from early development with a wide functional range.

PREVALENCE

~1 in 36 (US 8-year-olds)

TYPICAL ONSET

Signs by 12-24 months

SEX RATIO

~3.8:1 male:female

COURSE

Lifelong; ~1/3 minimally verbal

CLINICAL PICTURE

- Reduced social reciprocity with limited joint attention, atypical eye contact, and difficulty reading tone, gesture, and facial expression.
- Language ranges from fluent but pedantic and monotone speech to minimal verbal output; echolalia and scripted phrases are common.
- Insistence on sameness produces distress at transitions, rigid routines, and narrow special interests pursued with unusual intensity.
- Stereotyped motor movements such as hand flapping, rocking, spinning, and toe walking often intensify with excitement or stress.
- Sensory hyperreactivity or hyporeactivity to sound, texture, light, and pain drives much observable avoidance and meltdown behavior.
- Camouflaging is common in girls and adults, delaying diagnosis until anxiety, depression, or burnout finally brings them to care.

DIAGNOSTIC CRITERIA AT A GLANCE

- Requires all three social communication deficits: social-emotional reciprocity, nonverbal communication, and developing or maintaining relationships.
- Requires at least two of four restricted or repetitive domains: stereotypy, insistence on sameness, fixated interests, and sensory reactivity.
- Symptoms must be present in early development, though impairment may surface only when social demands exceed limited compensatory capacity.
- Severity is rated separately for social communication and for restricted behavior as level 1, 2, or 3 by amount of support required.
- Specifiers cover intellectual impairment, language impairment, associated medical, genetic, or environmental conditions, and catatonia.

NEUROBIOLOGY & PHYSIOLOGY

- Heritability estimates of **60-90%**; hundreds of risk genes converge on synaptic scaffolding, chromatin remodeling, and transcriptional regulation.
- Syndromic causes include fragile X, tuberous sclerosis, Rett syndrome, and 15q11-13 duplication; chromosomal microarray is first-tier testing.
- Early brain overgrowth across the first 2-3 years is followed by atypical cortical thinning and reduced long-range functional connectivity.
- Excitatory-inhibitory imbalance arising from GABAergic interneuron dysfunction contributes to sensory overload and elevated seizure risk.
- Amygdala and fusiform face area show atypical activation to faces, and superior temporal sulcus underresponds to biological motion.
- Advanced parental age, extreme prematurity, and in utero valproate exposure raise risk; vaccines have been definitively excluded as a cause.

PSYCHOLOGICAL MECHANISMS

- Theory of mind delay impairs inference of others' beliefs and intentions, producing literal, rule-bound interpretation of social situations.
- Weak central coherence biases processing toward detail over gestalt, aiding pattern detection but hindering use of context and meaning.
- Executive dysfunction in set shifting and planning underlies rigidity, transition distress, and difficulty coping with novel demands.
- Predictive coding accounts describe intolerance of prediction error, making unpredictable social and sensory environments intensely aversive.
- Repetitive behavior functions to regulate arousal; suppressing it without offering a replacement raises distress rather than reducing it.

DIFFERENTIAL & COMORBIDITY

- Distinguish from social (pragmatic) communication disorder, which lacks restricted repetitive behavior, and from selective mutism and social anxiety.
- Global developmental delay, severe early deprivation, hearing loss, and developmental language disorder can all mimic core social deficits.
- ADHD** co-occurs in 30-60%, anxiety disorders in about 40%, intellectual disability in roughly 35%, and epilepsy in 10-30%.
- Gastrointestinal symptoms, sleep-onset insomnia, and avoidant/restrictive food intake are common and often drive the presenting complaint.
- Elopement, self-injurious behavior, and elevated suicide risk in verbally fluent adults require explicit and repeated safety planning.

Treatment EVIDENCE-BASED MANAGEMENT

PHARMACOLOGIC

- No medication treats core social communication deficits; agents target irritability, aggression, hyperactivity, and repetitive behavior.
- Risperidone 0.5-3 mg/day** and **aripiprazole 2-15 mg/day** carry FDA approval for irritability associated with autism in ages 5-17.
- Monitor weight, BMI percentile, lipids, glucose, prolactin, and extrapyramidal signs at baseline, at 3 months, then at least annually.
- Methylphenidate** helps co-occurring **ADHD** but with lower response rates and more adverse effects than in ADHD without autism.
- Melatonin 1-6 mg** at bedtime improves sleep onset; SSRIs show weak evidence for repetitive behavior and frequently cause activation.

PSYCHOTHERAPY

- Early intensive behavioral intervention using **ABA** methods, 20-40 hours weekly for 1-3 years, has the strongest outcome evidence.
- Naturalistic developmental behavioral interventions such as the **Early Start Denver Model** embed teaching within play and daily routines.
- Speech-language therapy targets pragmatics and, when spoken language is absent, augmentative and alternative communication systems.
- PEERS** social skills groups over 14-16 weeks improve peer engagement in adolescents and young adults who have fluent language.
- Adapted **CBT** using visual supports and concrete language reduces co-occurring anxiety in cognitively able children and adolescents.

ADJUNCT & SUPPORTIVE

- M-CHAT-R/F** screening at 18 and 24 months; **ADOS-2** with **ADI-R** and developmental history anchor diagnostic confirmation.
- Chromosomal microarray and fragile X testing are recommended first-tier genetic workup for every newly diagnosed individual.
- Occupational therapy addresses sensory regulation, self-care, and motor coordination; physical therapy when hypotonia limits mobility.
- Individualized education programs, structured teaching, and visual schedules translate treatment goals into the school day.
- Transition planning from age 14-16 covers vocational training, guardianship or supported decision making, and adult service linkage.

CLINICAL PEARLS

- Loss of language or social skills after age 2 warrants neurology referral, not reassurance.
- Ask what function a repetitive behavior serves before trying to extinguish it.
- Fluent speech does not rule out autism; camouflaging delays diagnosis in girls.

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NOTE

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