



Autism Spectrum Disorder Part I: Overview, Screening, Diagnosis and Treatment Planning

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Learning Objectives



At the end of this educational activity, participants should be able to:

1. Identify symptoms or behaviors that may alert health care professionals to the need for targeted autism spectrum disorder (ASD) screening;
2. Compare and contrast evidence-based ASD screening tools;
3. State when to perform ASD screening to identify the need for subspecialty referral for evaluation; and
4. Explain the importance of people-first language.



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2

What you really need to know



Understand the special needs of children with ASD, in order to best support them, their caregivers and their families.

How to recognize a person with ASD – their behaviors and their medical needs.



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3

Sensory Overload – The world according to people with ASD



<https://www.youtube.com/watch?v=HzaBmwhTv6M>

The video was created by www.interactingwithautism.com and animator Miguel Jiro.

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4

What is Autism?

What is Autism?



Autism is a complex neurobiological disorder

- Onset before 3 years of age
- A range of abilities and disabilities
 - Intellectual disability
 - Social impairments
 - Types of unusual behaviors
 - Communication abilities
- Impairs and limits everyday functioning
- Lifelong disability



DSM-IV – Pervasive Developmental Disorders



- Autism
- Asperger Disorder
- PDD-NOS
- Child Disintegrative Disorder
- Rett Syndrome



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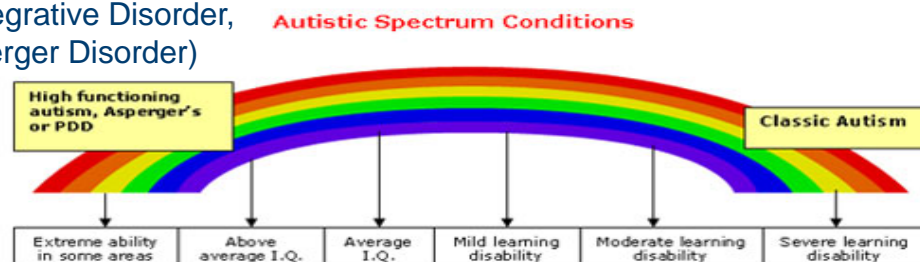
7

What is Autism Spectrum Disorder? DSM-5 Diagnostic Criteria



No more Rett Syndrome

ASD = all people on the spectrum
(including people previously
diagnosed with PDD-NOS,
Childhood Disintegrative Disorder,
Autism, and Asperger Disorder)



<https://www.dealwithautism.com/types-of-autism/>

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8

DSM-5 Criteria for ASD



- Deficits in social communication and social interaction
 - **Must meet all 3 criteria**
- Restrictive, repetitive patterns of behavior, interests or activities
 - **Meets 2 of 4 criteria**
- Must be present in the early developmental period
- Impair function
- Not otherwise explained by other developmental problems
- Severity levels

Deficits in social communication and social interaction



1. Deficits in social-emotional reciprocity
 - Unaware of other people's feelings
 - Problems identifying / understanding their own feelings
 - Lack of joint attention / reduced sharing of interests, emotions, and facial affect
 - Abnormal social approach / lack of initiation of social interaction
 - Failure of normal back-and-forth conversation
 - May not understand jokes or sarcasm
 - May not respond to his / her name

Deficits in social communication and social interaction



2. Deficits in nonverbal communicative behaviors used for social interaction



- Poor eye contact
- No social smile / lack of facial expression
- Deficits in understanding and use of nonverbal communication
- Does not point or gesture to communicate or share interest in something
- Poorly integrated verbal and nonverbal communication



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11

Deficits in social communication and social interaction



3. Problems developing and maintaining relationships, appropriate to developmental level (beyond those with caregivers)

- Difficulties adjusting behavior to suit different social contexts
- Difficulties in sharing imaginative play
- Problems initiating interactions, responding to others, sustaining interactions
- Prefers to be alone
- Absence of interest in people



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12

Restricted, repetitive patterns of behavior, interests, or activities



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

- Echolalia – immediate and delayed
- Idiosyncratic phrases
- Hand flapping, spinning, toe-walking
- Repetitive use of objects



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Restricted, repetitive patterns of behavior, interests, or activities



2. Excessive adherence to routines, ritualized patterns of verbal or nonverbal behavior, or excessive resistance to change

- Motoric rituals
- Insistence on same route or food
- Repetitive questioning
- Extreme distress at small changes

Restricted, repetitive patterns of behavior, interests, or activities



3. Highly restricted, fixated interests that are abnormal in intensity or focus

- Restricted interests with a specific toy (or part of a toy), hobby, or topic
- Strong attachment or preoccupation with unusual objects
- Excessively circumscribed or perseverative interests



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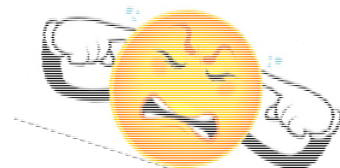
15

Restricted, repetitive patterns of behavior, interests, or activities



4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment

- Hypersensitive to loud noises, hair-washing, nail-cutting, tags, bright lights, food textures, chaotic environments, smells
- Sensory-seeking – licking, jumping, deep pressure
- Excessive smelling or touching objects
- Fascination with lights or spinning objects
- Indifference to pain / heat / cold



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16

Prevalence of ASD

Prevalence



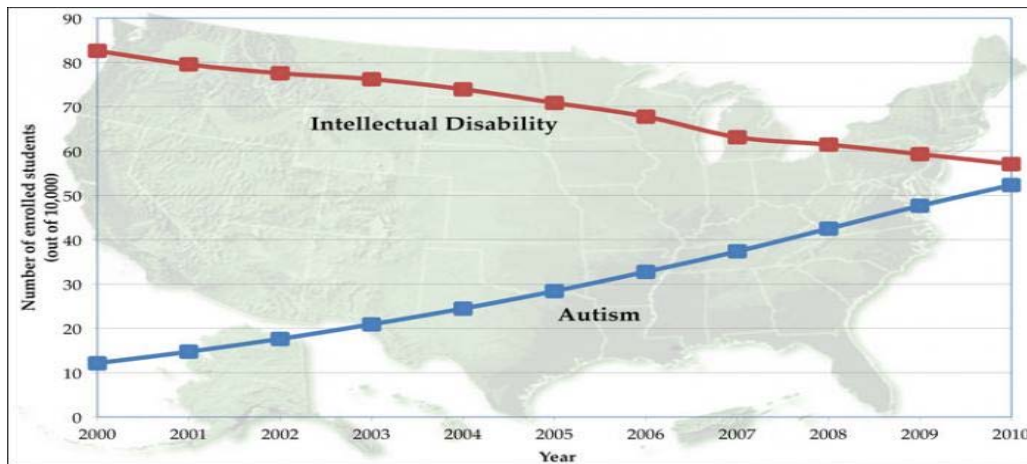
- **1 in 59** children in the U.S. / 16.8 per 1000, aged 8 years
 - **Up to 3 million** people in the US
- Occurs in all racial, ethnic, and socioeconomic groups
- **4 times more common in boys** (1 in 38) than among girls (1 in 151)



Autism and Developmental Disabilities Monitoring Network, CDC, 2014

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Prevalence: Change over time



www.autismspeaks.org

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Developmental and ASD Screening

Developmental Screening



- **1 in 4 children**, ages 0-5, are at moderate or high risk for developmental, behavioral, or social delay.
- ***Birth to 5: Watch Me Thrive!*** seeks to:
 - Celebrate milestones.
 - Promote universal screening.
 - Identify possible delays and concerns early.
 - Enhance developmental supports.



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Developmental Screening



AAP recommends:

- Developmental surveillance at every health supervision visit
- Developmental screening at 9, 18, and 30 months
- Or if parent or clinician has concerns

96110 - Billing code to use for developmental screen



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Developmental Screening



Dev Screening Tool	Ages served
Ages and Stages Questionnaire (ASQ-3)	1 - 66 months
Brigance screens	Birth to 1 st grade
Developmental Assessment of Young Children, 2 nd edition (DAYC-2)	Birth to 5 years
Early Screening Profiles	2 years – 6 years 11 months
Parent's Evaluation of Developmental Status (PEDS)	Birth to 8 years



Autism Screening



AAP recommends using a standardized ASD screen:

- At 18 and 24 months
- Or if parent or clinician has concerns

96110 - Billing code to use for developmental screen



Autism Screening



ASD Screening Tools	Ages Served
Ages & Stages Questionnaires SE-2	1 – 72 months
Childhood Autism Spectrum Test (CAST)	5 – 11 years
Communication and Symbolic Behavior Scales (CSBS DP)	6 - 24 months
M-CHAT-R	16 - 48 months
Pervasive Developmental Disorders Screening Test-II (PDDST-II)	12 – 48 months
Social Communication Questionnaire (SCQ)	4 years old and older



25

M-CHAT-R/F (2009)

- Parents answer 20 “yes/no” questions
- Score abnormal responses:
 - all answers should be “yes” except items 2, 5, and 12 (these are supposed to be no’s)
- Add up the abnormal responses

M-CHAT-R™

Please answer these questions about your child. Keep in mind how your child usually behaves. If you have seen your child do the behavior a few times, but he or she does not usually do it, then please answer no. Please circle yes or no for every question. Thank you very much.

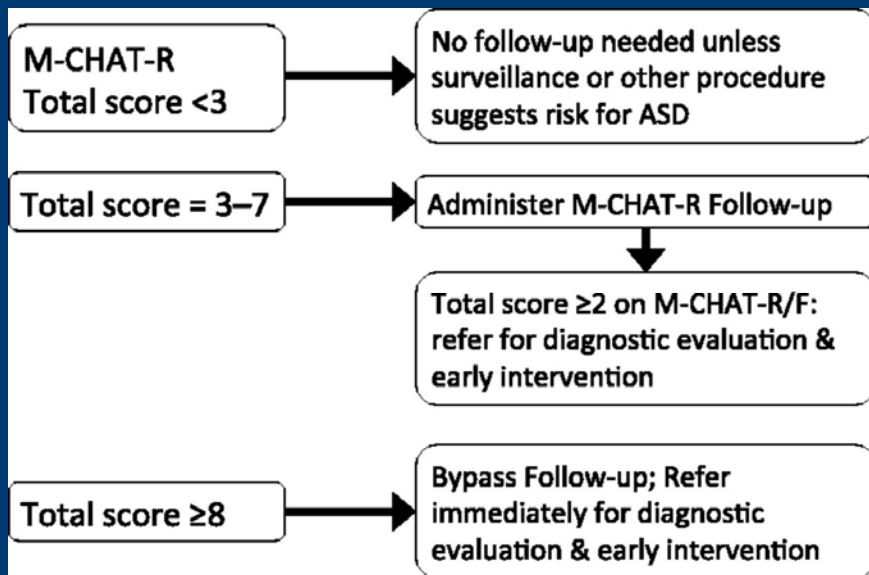
1. If you point at something across the room, does your child look at it? (FOR EXAMPLE, if you point at a toy or an animal, does your child look at the toy or animal?)	Yes	No
2. Have you ever wondered if your child might be deaf?	Yes	No
3. Does your child play pretend or make-believe? (FOR EXAMPLE, pretend to drink from an empty cup, pretend to talk on a phone, or pretend to feed a doll or stuffed animal?)	Yes	No
4. Does your child like climbing on things? (FOR EXAMPLE, furniture, playground equipment, or stairs)	Yes	No
5. Does your child make <u>unusual</u> finger movements near his or her eyes? (FOR EXAMPLE, does your child wiggle his or her fingers close to his or her eyes?)	Yes	No
6. Does your child point with one finger to ask for something or to get help? (FOR EXAMPLE, pointing to a snack or toy that is out of reach)	Yes	No
7. Does your child point with one finger to show you something interesting? (FOR EXAMPLE, pointing to an airplane in the sky or a big truck in the road)	Yes	No
8. Is your child interested in other children? (FOR EXAMPLE, does your child watch other children, smile at them, or go to them?)	Yes	No
9. Does your child show you things by bringing them to you or holding them up for you to see – not to get help, but just to share? (FOR EXAMPLE, showing you a flower, a stuffed animal, or a toy truck)	Yes	No
10. Does your child respond when you call his or her name? (FOR EXAMPLE, does he or she look up, talk or babble, or stop what he or she is doing when you call his or her name?)	Yes	No
11. When you smile at your child, does he or she smile back at you?	Yes	No
12. Does your child get upset by everyday noises? (FOR EXAMPLE, does your child scream or cry to noise such as a vacuum cleaner or loud music?)	Yes	No
13. Does your child walk?	Yes	No
14. Does your child look you in the eye when you are talking to him or her, playing with him or her, or dressing him or her?	Yes	No
15. Does your child try to copy what you do? (FOR EXAMPLE, wave bye-bye, clap, or make a funny noise when you do)	Yes	No
16. If you turn your head to look at something, does your child look around to see what you are looking at?	Yes	No
17. Does your child try to get you to watch him or her? (FOR EXAMPLE, does your child look at you for praise, or say “look” or “watch me?”)	Yes	No
18. Does your child understand when you tell him or her to do something? (FOR EXAMPLE, if you don’t point, can your child understand “put the book on the chair” or “bring me the blanket?”)	Yes	No
19. If something new happens, does your child look at your face to see how you feel about it? (FOR EXAMPLE, if he or she hears a strange or funny noise, or sees a new toy, will he or she look at your face?)	Yes	No
20. Does your child like movement activities? (FOR EXAMPLE, being swung or bounced on your knee)	Yes	No



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26



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After Diagnosis, What next?

PCP - Initial Assessment



History

- Prenatal and postnatal
- Ask about seizures, sleep, nutrition / GI

Hearing / Vision

Physical / Neurological examination

- Growth, Head Circumference
- Dysmorphic features
- Neurocutaneous markings



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29

PCP - Initial Assessment



Referrals:

- Audiology / Ophthalmology
- Speech / Language
- Occupational therapy

Labs:

- Nutrition labs, lead, CBC, Ferritin, TSH
- Fragile X and CMA



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30

Genetic abnormalities assoc. with ASD

Syndrome	Implicated Gene (s)
2q37 deletion	Possibly CENTG2
15q11-q13 duplication	UBE3A, GABRB3, possibly others
16p11.2 deletion	Unknown
22q11.2 deletion	Unknown
22q13.3 deletion	SHANK3
Fragile X syndrome (locus Xq27.3; abnormal expansion of CGG-trinucleotide repeats)	FMR1
Macrocephaly / autism syndrome, PTEN hamartoma tumor syndrome (locus 10q23.31)	PTEN
Rett syndrome (locus Xq28)	MECP2
Tuberous Sclerosis (loci 9q34 and 16p13.3)	TSC1, TSC2



Developmental and Behavioral Pediatrics, AAP

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31

Medical Home

- A concept, not a place
- Family-Centered Care
- Family perspective:
 - Responsive system of services that should provide a seamless and transparent spectrum of services
 - Accessible, flexible, responsive
 - Targeted to address the child's mental, physical, emotional and social needs



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32

People First Language Essential for Family-Centered Care

A disability descriptor is simply a medical diagnosis.

People First Language respectfully puts the person before the disability.

A person with a disability is more *like* people without disabilities than different!

- **NOT** the Downs Kid – The child with Down syndrome
- **NOT** the Autistic Kid – The child with Autism
- **NOT** the Retarded Kid – The child with an intellectual disability



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In Summary Best Practice Recommendations



- Follow screening guidelines
- Identify early
- Provide Medical Home / Family-centered Care
 - Use People First Language
- **Collaboration** of providers and staff with family members and their caregivers is crucial

**Listen to parents' concerns.
Remember – Parents are the Experts!**



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Resources and References

References / Resources



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- Early Screening Profiles
 - <https://www.pearsonclinical.com/childhood/products/100000089/early-screening-profiles-esp.html>
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 - <http://www.pedstest.com/default.aspx>

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37

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- Ages and Stages Questionnaire – Social-Emotional
 - <http://agesandstages.com/products-services/asqse-2/>
- Social Communication Questionnaire
 - <http://www.wpspublish.com/store/p/2954/social-communication-questionnaire-scq>

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38

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 - http://www2.gsu.edu/~psydlr/M-CHAT/Official_M-CHAT_Website_files/M-CHAT-R_F.pdf
- CSBS DP Infant Toddler Checklist
 - [Firstwords.fsu.edu/pdf/checklist.pdf](http://firstwords.fsu.edu/pdf/checklist.pdf)
 - <https://firstwords.fsu.edu/checklist.html>
- Childhood Autism Screening Test (CAST)
 - www.autismresearchcentre.com/project_9_cast

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39

What's Next?

SNI / OHE Collaboration ASD Webinar Series



- Part II: Medical Home care
- Part III: Genetics and ASD
- Part IV: Treatment Strategies
- Part V: Dual Diagnosis of Down syndrome and ASD
- Part VI: Transition to Adult Care



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41

Thank you

Q&A