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Autism Spectrum Disorder Diagnosis in the General Pediatrics Setting

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Learning Objectives

- Understand growing prevalence of autism spectrum disorder and barriers to evaluation/diagnosis.
- Review components of an autism diagnostic evaluation per clinical practice guidelines.
- Review tools which may be utilized in the primary care setting for autism diagnosis.
  - CARS, STAT
- Understand documentation requirements of an autism diagnostic evaluation to ensure access to treatment.
- Review treatments for autism spectrum disorder, which may be prescribed by the primary care provider.

Autism's Increasing Prevalence

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Prevalence

- Before 1990: 1 out of 2,222 children
- March 2023: 1 out of 36 US children (2020 data set)
- 1 out of 34 children in NJ
- Male to female ratio: 4 to 1

Autism and Developmental Disabilities Monitoring Network, CDC

Why the increase?

- Change in diagnostic tendencies?
- Increased public awareness
- Shift to younger age of diagnosis
- Need for diagnostic label to obtain services
- True increase in incidence?
- Unidentified environmental factor
- Genetic factor

Autism: Race from diagnosis to intervention

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- Prompt diagnosis and initiation of treatment significantly improves outcomes[1].
- Waits for evaluation and diagnosis are "the norm."
  - Average delay of 2 years from developmental screening to autism diagnosis[2].

Autism: Race from diagnosis to intervention

- Jake is a 32 month old boy presenting Developmental Behavioral Pediatrics evaluation.
  - 18 month Well Check by his PCP- Developmental concerns; referred to Early Intervention and Audiology
  - 24 month Well Check, M-CHAT-R score was 15 (High Risk)- Referred to Developmental Pediatrics

- The hallway…
  - Jake shows no response to examiner's greeting or his name.
  - Hand-flapping and nonsensical verbalizations are observed.

- Mother asks if she can get "a diagnosis" today.
  - "He needs a diagnosis so he can get services."

Autism: Race from diagnosis to intervention

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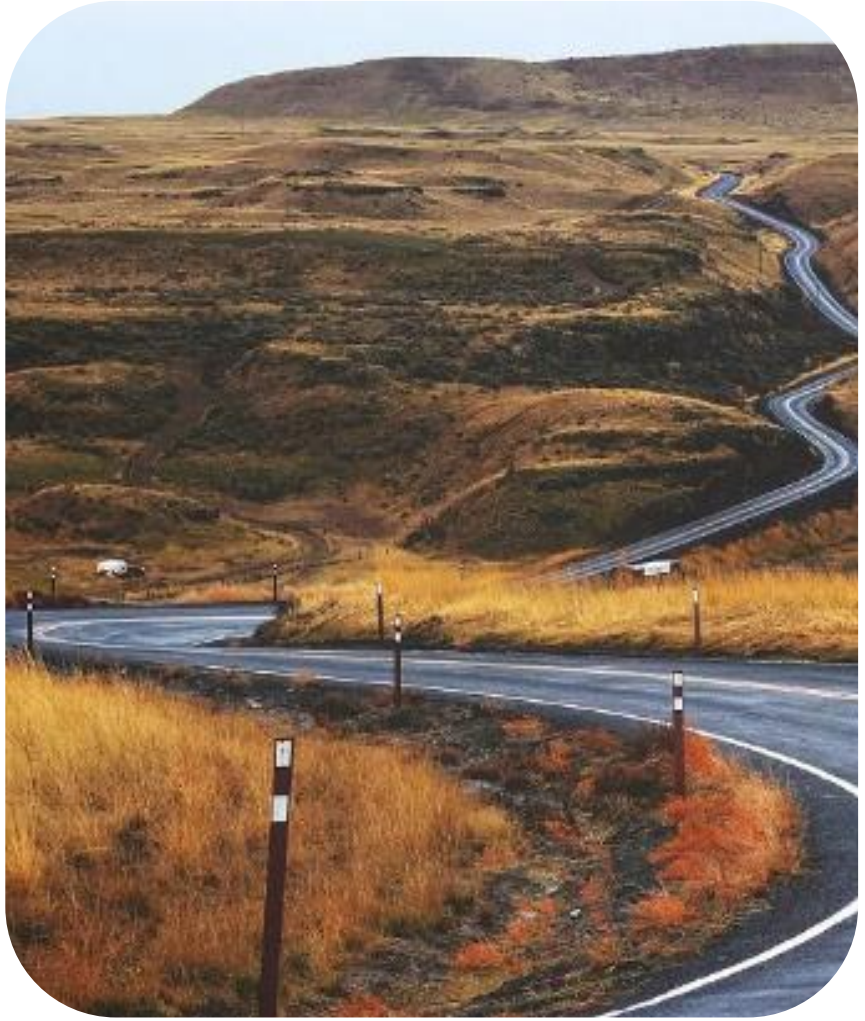
- Autism diagnosis and recommendations made for:
  - ✓ Applied Behavior Analysis (ABA)
  - ✓ Continuation of Early Intervention (ST, OT)
  - ✓ Transition to the District at 36 months
- Jake returns at <u>37 months</u> for follow up.
  - ✓ He has an IEP- receives ST, OT and a special education preschool program
  - ✓ Waitlisted with four ABA agencies
- Jake returns at <u>43 months</u> for follow up.
  - ✓ 15 hours/week of ABA services initiated last week
  - ✓ Approved for 20 hours but agency unable to fully staff



25 months passed between PCP and full services.

Barriers to Autism diagnosis:

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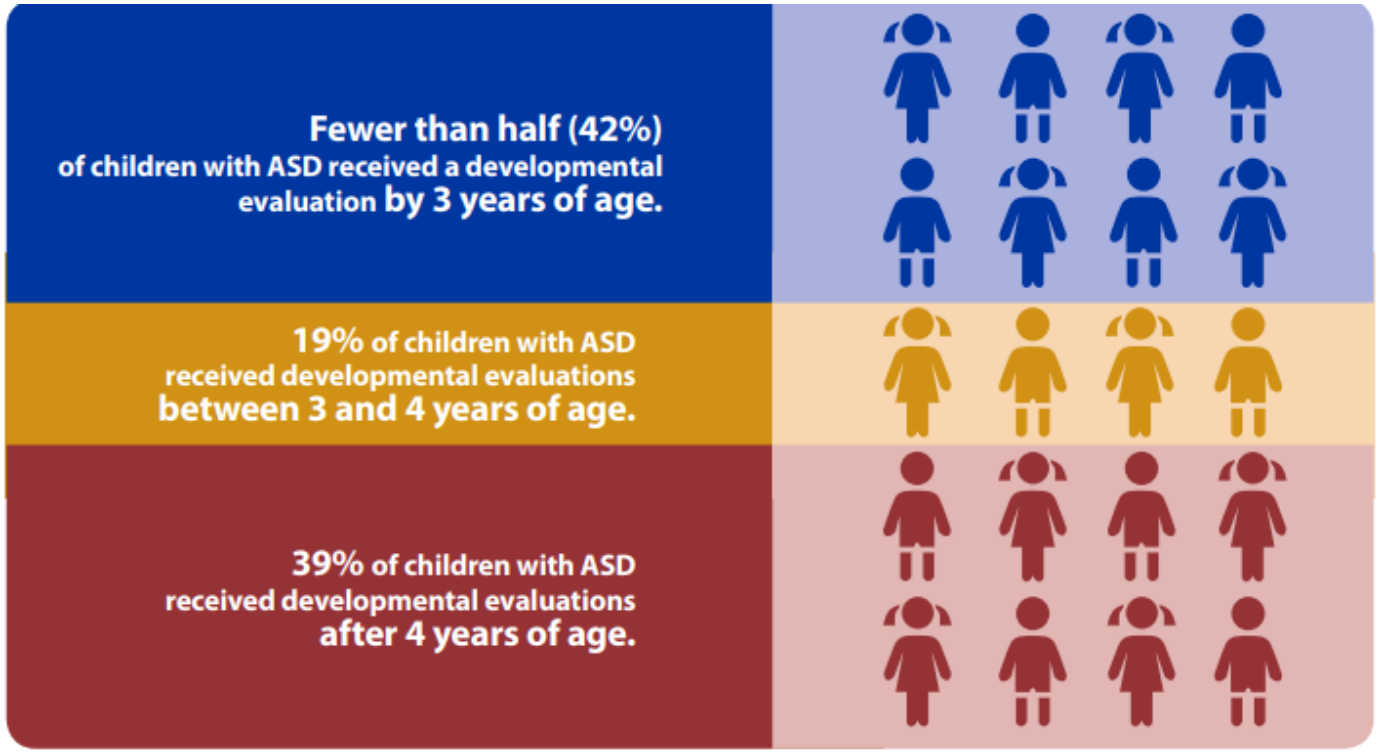
85% of children identified with ASD had concerns about their development noted in their medical records by 3 years of age.

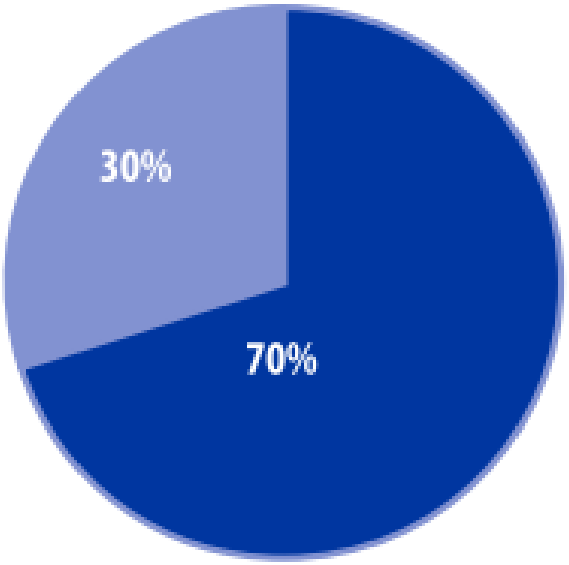


https://www.cdc.gov/ncbddd/autism/addm-community-report/documents/spotlight-on-delay-addm-community-report-2018-h.pdf

Barriers to Autism diagnosis:

Diagnosis

A **medical diagnosis** can be a key step in getting medical services provided through health insurance.



Of the children who received an ASD diagnosis, the average age of diagnosis was a little under 4 and a half years (4 years and 4 months).

30% of children who met the ADDM Network surveillance case criteria for ASD **had not received a formal ASD diagnosis by 8 years of age,** potentially limiting the services they receive.

https://www.cdc.gov/ncbddd/autism/addm-community-report/documents/spotlight-on-delay-addm-community-report-2018-h.pdf

Barriers to Autism diagnosis

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Disorder Posing Unique Diagnostic Challenges

- No biomarkers
- Heterogeneous, with range of severity and symptom expression
- Comorbidities

Barriers to Autism diagnosis

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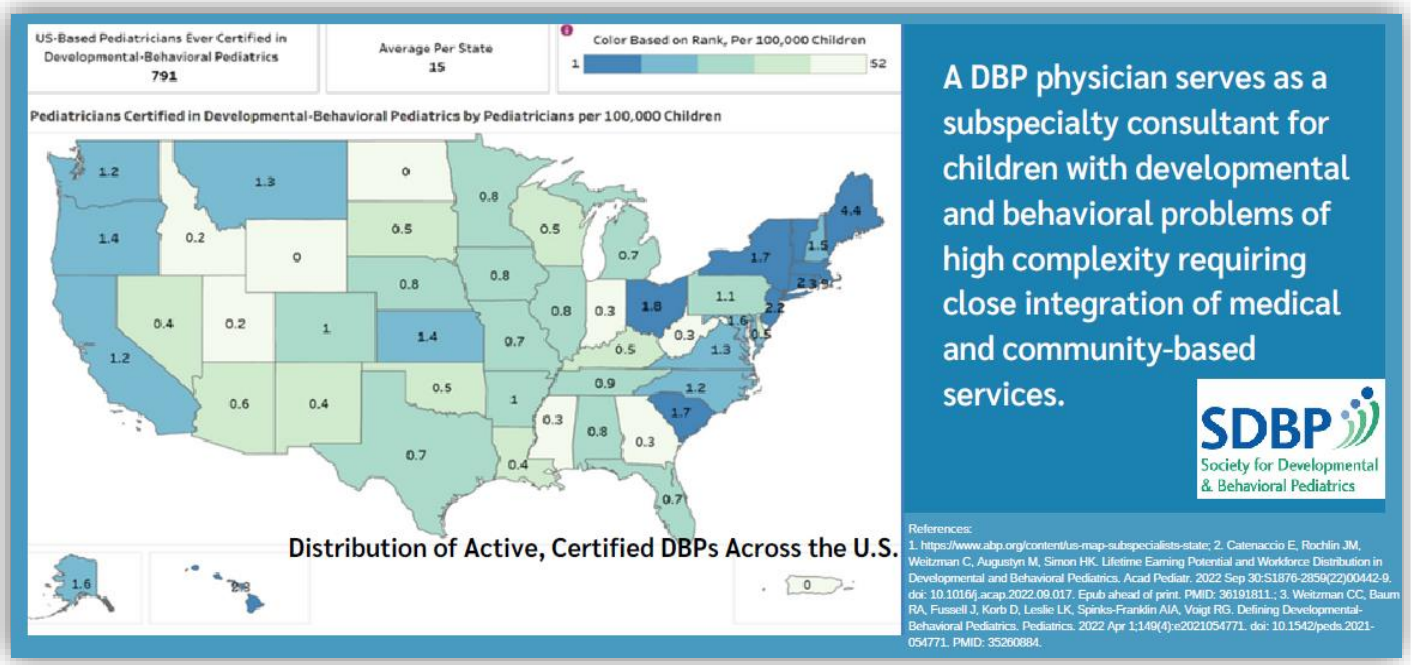
Confounding Variables

- Behavior may vary by context or relationship
- Different information sources may be contradictory
- Social and cultural variance –
  Impacts how information is reported or interpreted
  (socio-economic status, parental factors)

Barriers to Autism diagnosis:

- Subspecialist access
  - Child Neurology
  - Developmental Behavioral Pediatrics (DBP)
  - Child Psychiatry
- Shrinkage of DBP field
  - 706 physicians maintain certification in DBP
  - Many report retirement in the next 5 years



https://sdbp-video.s3.amazonaws.com/wp-content/uploads/2022/10/26151426/Value-DBP_INFOGRAPHIC_REV.pdf

How is Autism diagnosed?

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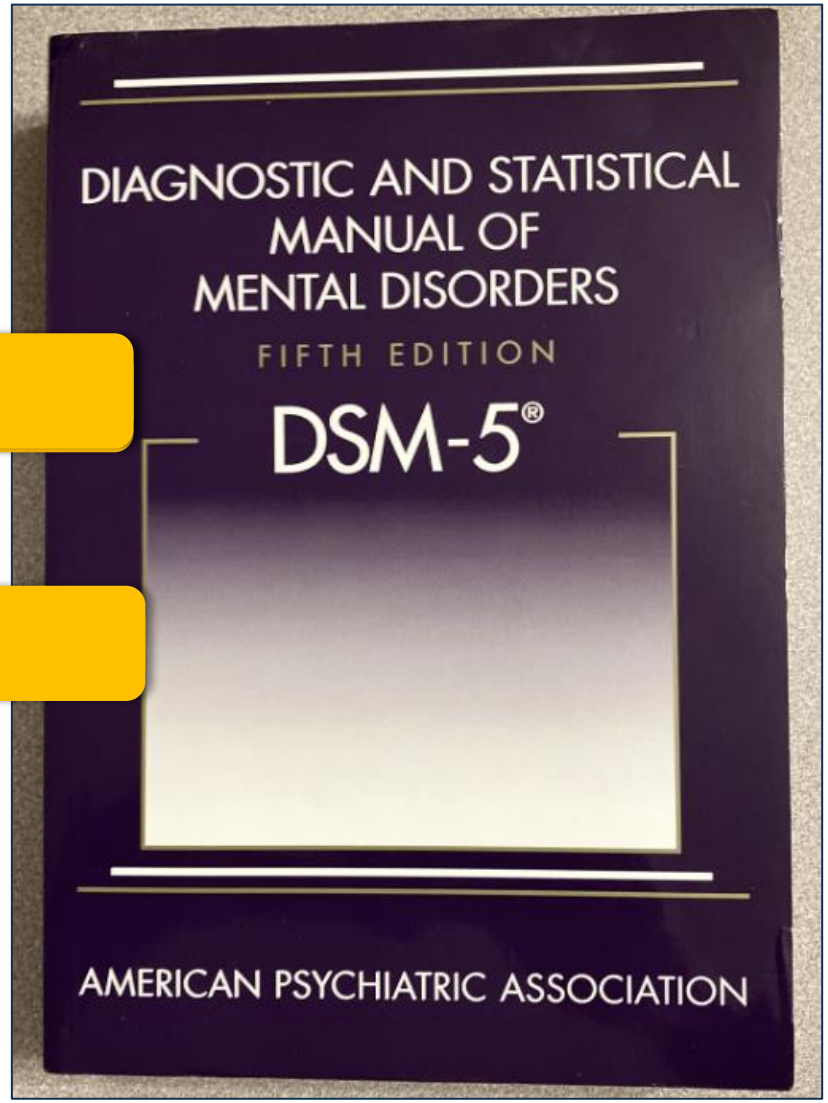
Establishment of DSM-5 criteria

Who can diagnose?

What is "required"?



DSM-5 Criteria for ASD

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:
   - Abnormal social approach and 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 communicative behaviors used for social interaction:
   - Poorly integrated verbal and nonverbal communication
   - Abnormalities in eye contact and body language or deficits in understanding and use of gestures
   - 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 in sharing imaginative play or in making friends
   - Absence of interest in peers

DSM-5 Criteria for ASD

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:
   - Motor stereotypies, lining up toys, echolalia.
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior:
   - Distress at small changes, difficulties with transitions, rigid thinking.
3. Highly restricted, fixated interests that are abnormal in intensity or focus:
   - Preoccupation with unusual objects, perseverative interest.
4. Hyper or hypo-reactivity to sensory input or unusual interests in sensory aspects of the environment.
   - Indifference to pain/temperature, adverse response.

DSM-5 Criteria for ASD

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

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

E. 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 that expected for general developmental level

Who can diagnose Autism?

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Current state: Subspecialist Evaluation

Two Parts to Assessment

- Clinical observation
- Review of history provided by multiple caregivers – parents, teachers, etc.

Evidence-based tools exist for both parts; use of these tools varies significantly

What is “required” to diagnose Autism?

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Many Clinical Practice Guidelines (CPGs) exist for ASD evaluation, but quality and content vary significantly

- Penner et al. completed a review of 869 published English language autism diagnostic guidelines in 2018.
- 11 were identified, meeting inclusion and quality criteria[3]:

Table 2. Title, year, and type of guidance documents used in review.

Guideline, year	Type of document	Abbreviation
American Academy of Neurology, 2000	Practice parameter	AAN
American Academy of Pediatrics, 2001	Policy statement	AAP 2001
British Columbia, 2003	Standards and guidelines	BC
American Speech-Language-Hearing Association, 2006	Guideline	ASHA
American Academy of Pediatrics, 2007	Clinical report	AAP 2007
Scottish Intercollegiate Guidelines Network, 2007	Guideline	SIGN
New Zealand Ministries of Health and Education, 2008	Guideline	NZ
Miriam Foundation, 2008	Best practice guidelines	Miriam
American Occupational Therapy Association, 2009	Practice guideline	AOTA
National Institute for Health and Care Excellence, 2011	Guideline	NICE
American Academy of Child and Adolescent Psychiatry, 2014	Practice parameter (patient-oriented)	AACAP

“Gold Standards” in ASD Evaluation

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Review of CPGs revealed the following “Gold Standard” components[3]

- Multi-disciplinary Team (MDT)
- Evidence-based Tools

Need for Flexibility with the MDT

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There is little evidence suggesting that a MDT assessment is more accurate than that of a solo clinician or more capable of developing a list of differential diagnoses[3]

When MDT May Not be Needed

- **Cases in which the diagnosis is "obvious"**
- Cases where the practitioner cannot easily access a MDT assessment[4]

Diagnostic Tools for ASD Evaluation

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Area of significant variation in practice

Three instruments with strongest supporting evidence[4]

- Autism Diagnostic Observation Schedule (ADOS)
- Autism Diagnostic Interview Revised (ADI-R)
- Childhood Autism Rating Scale (CARS)

Diagnostic Tools for ASD Evaluation

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Questions Raised[5]

Clinical benefits using these tools

Possibility of harm that could arise from relying solely on the score provided by the tool

Need for Flexibility with Tools

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AACAP[6]: Highlights importance of clinical judgment

"The use of such instruments supplements, but does not replace, informed clinical judgment"

CDC[7]

"There are many tools to assess ASD…but no single tool should be used as the basis for diagnosis…"

Need for Flexibility with Tools

Miriam Foundation[8]:
Considered the "gold standard for diagnosis…in Canada"

"a lack of ADI-R, ADOS data should not prevent a child from receiving much needed services if a diagnostician with sufficient expertise conducts the assessment."

Common themes…

- Current diagnostic practices are resource and time intensive.
- Flexibility in the diagnostic process is critical.
- Children with “obvious” presentations of autism may undergo a more streamlined assessment.
- General pediatricians may serve an important role in expansion of diagnostic access.

“Expanding the diagnostic workforce can facilitate timelier access to diagnosis and intervention; however, these assessments must be accurate.[9]”

Accuracy of General Pediatrician Diagnosis

- 17 general pediatricians (GP) and 106 children with possible ASD
- When the GP thought ASD was present, the MDT agreed 89% of the time
- When the GP did not think ASD was present, the MDT agreed only 60% of the time

- Penner M, Senman L, Andoni L, et al. Concordance of Diagnosis of Autism Spectrum Disorder Made by Pediatricians vs a Multidisciplinary Specialist Team. JAMA Netw Open. 2023;6(1):e2252879. doi:10.1001/jamanetworkopen.2022.52879

"…general pediatricians have a very high likelihood of correctly diagnosing ASD in children, particularly when they feel certain."

Table 3. Accuracy Results, Certainty, and Out-of-Study Pediatrician Actions

Variable	Cases, No. (%)			
	MDT diagnosis of ASD		MDT diagnosis not ASD	
	Pediatrician diagnosis correct (n = 54)	Pediatrician diagnosis incorrect (n = 18)	Pediatrician diagnosis correct (n = 27)	Pediatrician diagnosis incorrect (n = 7)
Pediatrician certainty, Likert scale				
1	0	0	0	0
2	1 (2)	3 (17)	1 (4)	2 (29)
3	10 (19)	7 (39)	11 (41)	3 (43)
4	19 (35)	7 (39)	13 (48)	1 (14)
5	24 (44)	1 (6)	2 (7)	1 (14)
MDT certainty, Likert scale				
1	0	0	0	0
2	0	0	3 (11)	0
3	2 (4)	3 (17)	9 (33)	3 (43)
4	11 (20)	7 (39)	14 (52)	2 (29)
5	41 (76)	8 (44)	1 (4)	2 (29)
Pediatrician action				
Tell family child has ASD	37 (69)	0	0	1 (14)
Tell family child does not have ASD	0	8 (44)	12 (44)	0
Refer to subspecialist	14 (26)	7 (39)	3 (11)	5 (71)
Watch and wait	3 (6)	2 (11)	4 (15)	0
Tell family child has different diagnosis	0	1 (6)	8 (30)	1 (14)

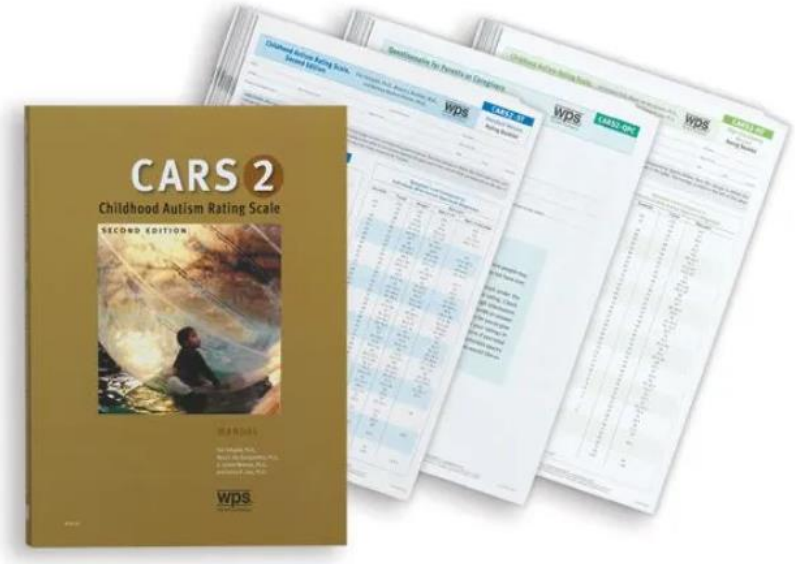
Factors that increase certainty…

- Parent concern[10]
- Therapist/teacher concern
- Family history of autism spectrum disorder[7]
- High risk on screening tool (MCHAT-R)
- Access to additional tools to:
  1. Elicit behavioral observations
  2. Obtain behavioral history from caregivers

Tools to aide Autism diagnosis

- Screening Tool for Autism in Toddlers & Young Children (STAT)
  - Interactive measure, to support behavioral observation
  - STAT™ | STAT (vueinnovations.com)

- Childhood Autism Rating Scale , 2nd Edition (CARS-2)
  - Clinician completed rating scale
  - (CARS™2) Childhood Autism Rating Scale™, Second Edition (wpspublish.com)



Screening Tool for Autism in Toddlers & Young Children (STAT)

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Training on the STAT can be accomplished in three ways, depending on your professional needs and background.

STAT™ Training Workshop

A small group setting for learning and practicing STAT administration and scoring.

Online STAT™ Training Tutorial

For those who prefer to learn the STAT at their own pace.
Learn More

STAT-MD Training

For physicians in pediatric practices seeking a more comprehensive approach to early identification that combines the STAT with other techniques.

- Developed to screen for autism in children between 24 and 36 months of age
- Designed for use by community service providers who work with young children
- Consists of 12 items, takes about 20 minutes to administer
- Assesses key social and communicative behaviors including imitation, play, requesting, and directing attention

Childhood Autism Rating Scale , 2nd Edition (CARS-2)

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2 Rating Scales

- 15 rating areas with a 7 point rating system, takes 5–10 minutes after the information needed to make the ratings has been collected)
- Rating areas based on central features and behavioral characteristics of ASD
- Ratings based on direct behavioral observations and information from multiple sources

CARS2-ST

- ✓ 24 months to under 6 years of age
- ✓ Over age 6 with:
  - IQ of 79 or below
  - Notably impaired communication

CARS2-HF

- ✓ 6 years of age and older
- ✓ IQ of 80 or higher
- ✓ Fluent communication

Autism: Race from diagnosis to intervention

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Purpose of diagnosis = Treatment

Payment for treatment is critical

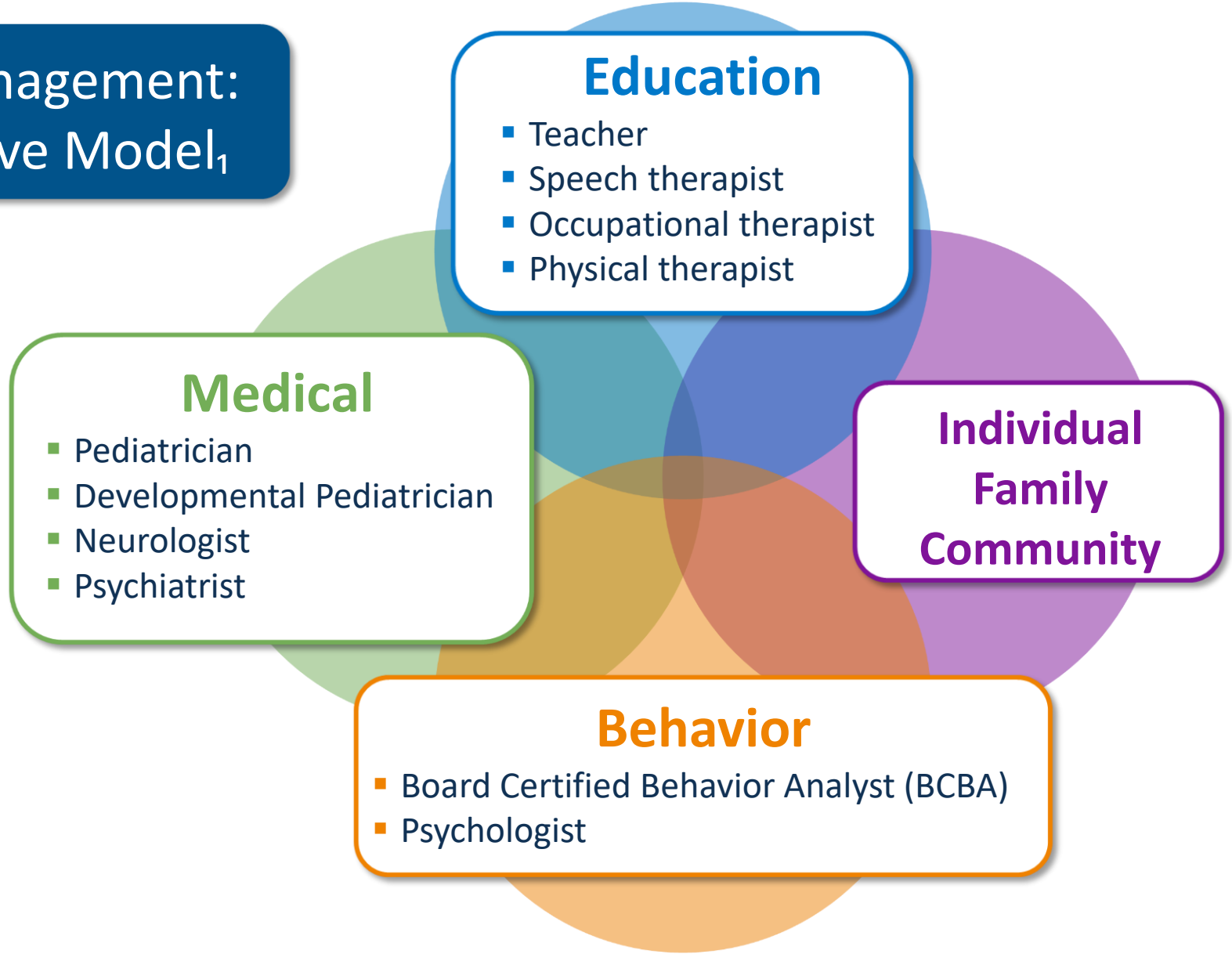
Documentation must meet insurance standards

Documentation Requirements

Applied Behavior Analysis (ABA) Comprehensive Diagnosis Checklist

- Signed on the bottom by a MD/DO
- Current within the last 5 years
- Includes diagnostic code of F84.0
- Includes the medical and developmental history of person
- Includes a narrative summary of how symptoms of a person fit DSM-5 criteria
- Includes one or more of the following assessments:
  - Autism Behavior Checklist [ABC]
  - Childhood Autism Rating Scale [CARS]
  - Autism Screening Questionnaire [ASQ]
  - Autism Quotient [AQ]
  - Autism Diagnostic Interview-Revised [ADI]
  - Autism Diagnostic Observation Schedule [ADOS] 1-2 edition
  - Gilliam Autism Rating Scale [GARS} 1-2

Autism Management: Collaborative Model[1]



Autism Management

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ASD is not "curable" and should be viewed as a chronic condition requiring management[1].

Goals of Treatment

Minimize core features
- Impairment in social reciprocity
- Deficits in communication
- Restricted, repetitive behaviors

Improve quality of life

Alleviate stress

Neurodiversity

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Diversity of human brains and minds – the infinite variation in neurocognitive functioning within our species

- Natural and valuable form of human diversity
- There is no "normal" or "healthy" type of brain or mind, or one "right" style of neurocognitive functioning

Equal access, rights, and opportunities

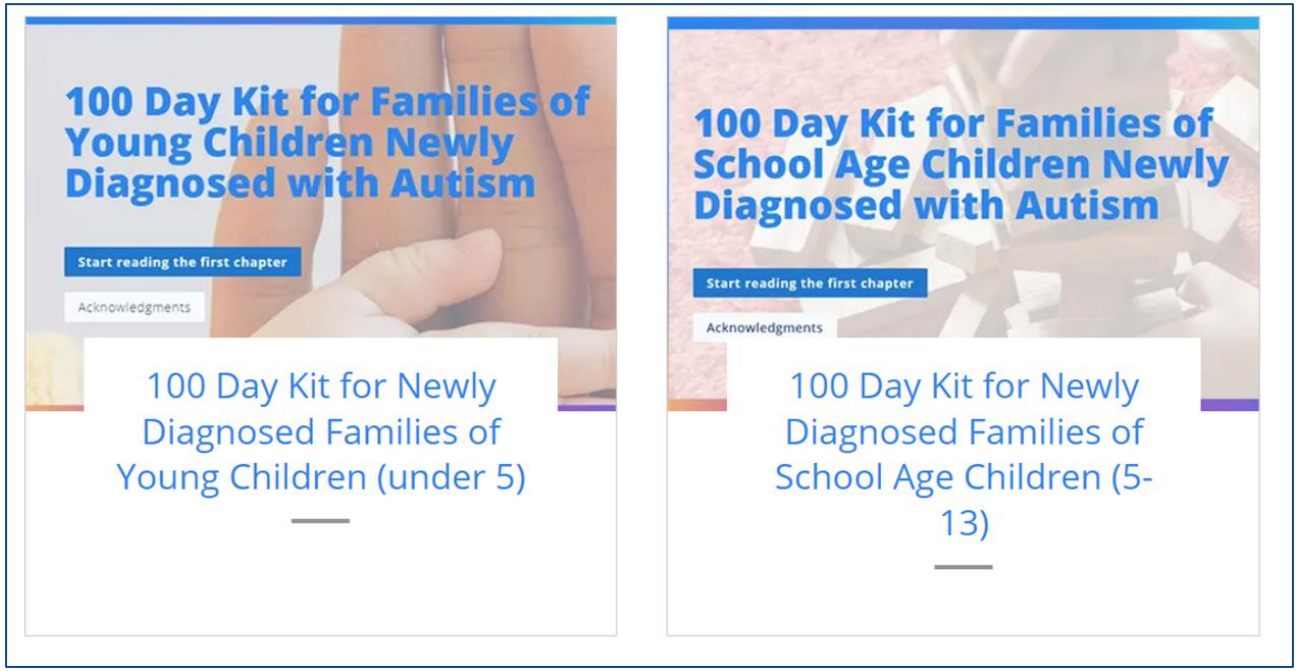
Self-determination and choice

Advocacy

http://autisticadvocacy.org/
http://neurocosmopolitanism.com/neurodiversity-some-basic-terms-definitions/

Autism Management

- Educational services
  - ✓ Early Intervention
  - ✓ Child Study Team
- Community/private therapies
  - ✓ Speech Therapy
  - ✓ Occupational Therapy
- Behavioral therapy
  - ✓ Applied Behavior Analysis (ABA)
  - ✓ Denver Model
  - ✓ Floor Time
- Specialist referral to support on-going care needs



Applied Behavior Analysis (ABA)

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The Science of Human Behavior

"Applied Behavior Analysis is the process of systematically applying interventions based upon the principles of learning theory to improve socially significant behaviors to a meaningful degree, and to **demonstrate that the interventions employed are responsible for the improvement** in behavior"

Substantial gains demonstrated in the following

- IQ
- Language
- Academic performance
- Adaptive behavior as well as some measures of social behavior

Outcomes significantly better than those of children in control groups

Baer DM, Wolf MM, Risley TR. Some current dimensions of applied behavior analysis. *Journal of Applied Behavior Analysis. 1968;1, 91-97.*

Autism Management

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Medication may be used to modify specific maladaptive behaviors[1]

- Repetitive, obsessive compulsive
- Anxiety
- Hyperactivity, impulsivity
- Inattention
- Aggression
- Self-injury
- Mood lability
- Depression

There is no medication "for "ASD"

Conclusions

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- Access to pediatrics subspecialists for autism evaluation is insufficient, resulting in delays in diagnosis.
- Early initiation of autism interventions (ST, OT, ABA, etc) results in improved outcomes. Diagnosis may be required for service initiation.
- The diagnosis of autism requires demonstration of DSM-5 criteria- flexibility is key when considering provider type or tool used for the process.
- General Pediatricians can accurately diagnose autism in cases where they have a high level of suspicion.
- Use of "gold standard" tools will aid the diagnostic process, and decrease likelihood of service denials by insurance.

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Thank you.

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