Autism: Screening, Diagnosis, and Beyond!

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Learning Objectives

1. Name 2 screening measures & 2 diagnostic measures
2. Explain the importance of prospectively tracking social communication skills in infancy and toddlerhood
3. Identify several evidence-based interventions frequently used with autistic individuals

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When I Think of Autism...

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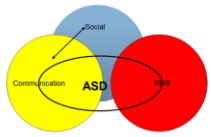
What are the core components of autism?

Autism is a developmental disorder characterized, to varying degrees, by difficulties in the areas of

- social interaction,
- verbal & nonverbal communication, &
- restricted & repetitive behaviors.

Core Symptoms



Based on Kaufman, W. (2012). The New Diagnostic Criteria for Autism Spectrum Disorders

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Autism Spectrum Disorder (ASD): Quick Facts

1:36 prevalence rate. Was 1:150 in the year 2000.

% identified varies by location, from a low of 1 in 60 (1.7%) children in Missouri to a high of 1 in 26 (3.9%) children in California (ADDM Network 2021)

4 autistic boys for every 1 girl with autism

Starting with last year's ADDM data, no difference in the number of African American children identified with autism compared to Caucasian children.

However, the number of Hispanic children identified with autism is still comparatively lower.

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Autism Spectrum Disorder (ASD): Quick Facts

- Some core symptomology needs to be present in very early childhood, but varied presentation
- Stable disorder. There is no "cure," but there are evidence-based interventions targeting specific skills.
- Heritable (No single cause; genetic & environmental contributions)
- Co-occurring conditions are common (e.g., 1/3 have co-occurring Intellectual Disability, ADDM Network 2020)
- On an individual basis, the lifetime cost of an autism spectrum disorder ranged from $1.43 million to $2.44 million, again depending on the presence or absence of an intellectual disability

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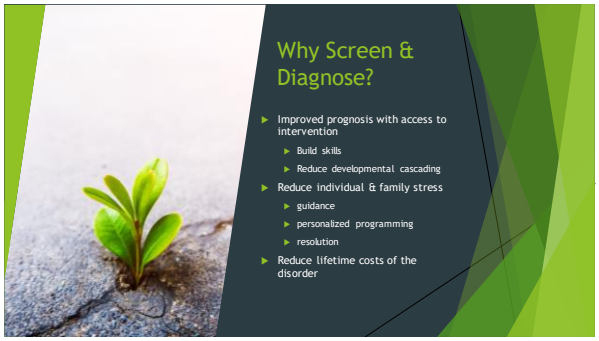
Why Screen & Diagnose?
Improved prognosis with access to intervention
Build skills
Reduce developmental cascading
Reduce individual & family stress
guidance
personalized programming
resolution
Reduce lifetime costs of the disorder

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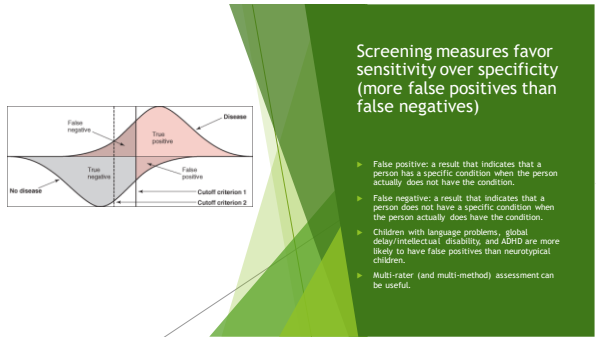
Screening measures favor sensitivity over specificity (more false positives than false negatives)
False negative
True positive
Disease
True negative
False positive
No disease
Cutoff criterion 1
Cutoff criterion 2
False positive: a result that indicates that a person has a specific condition when the person actually does not have the condition.
False negative: a result that indicates that a person does not have a specific condition when the person actually does have the condition.
Children with language problems, global delay/intellectual disability, and ADHD are more likely to have false positives than neurotypical children.
Multi-rater (and multi-method) assessment can be useful.

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AAP Developmental Screening Guidelines
The American Academy of Pediatrics recommends developmental screening at well-child visits.
All children screened to assess their general development at 9,18 and 24 or 30 months
All children screened for autism at 18 & 24 months (i.e., Level 1 screening)

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Level 1 Screening

- Modified Checklist for Autism in Toddlers, Revised with Follow-Up (M-CHAT-R/F)
  - For all children 16-30 months
  - If the M-CHAT-R results are high-risk (scores of 8-20), then child has screened positive
  - If the M-CHAT-R results are medium-risk (scores of 3-7), then ask "F" questions (follow up questions) before proceeding. If M-CHAT-R/F score remains at or above 2, the child has screened positive.
- Options for older children include the SRS-2 and the SCQ.
- Using the M-CHAT-R/F with children older than 30 months, will result in more false negatives.

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Level 2 Screening

Screening Tool for Autism in Toddlers (STAT)

- For children 14-47 months of age at risk for ASD based a positive M-CHAT-R/F, a positive SRS-2, and/or professional judgment.
- 12 items directly assessing key social and communication skills (e.g., imitation, play, requesting).
- Best research evidence is for use between the ages of 24-36 months of age.
- If the child scores at or above the cut-off on STAT - refer for further diagnostic evaluation.

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Level 2 Screening

Rapid Interactive Screening Test for Autism (RITA-T) in Toddlers

- Reliable in 18-36 months old toddlers
- Easy to train and learn; 5-10 minutes to administer and score
- Can be administered after a positive Level 1 test to identify those with real ASD risk
- The RITA-T differentiates extremely well between toddlers with ASD and toddlers with Developmental Delay (DD) /Non-ASD
- Toddlers with no apparent developmental delay and toddlers with developmental or language delays, but with intact social communication skills, score comparably on the RITA-T
- The RITA-T correlates well with established diagnostic measures of ASD.

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Prospective Approaches to Early Detection

- The addition of prospective research designs (e.g., infant sibling studies) has provided new information about what we can see and not see during the first two years of life.
- Before infant sibling studies, there were retrospective studies (e.g., parent interview, medical record reviews, home movies)
  - What did we record?
  - Who can remember?
    - "Forward telescoping"

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Infant Sibling Studies

- Ozonoff & Iosif, 2019
  - Social communication regression is common, not rare.
  - It can be observed over time (6-36 months)
  - It does not correspondence well to what people remember.
  - The declining trajectory may start even earlier than what we can detect through behavioral observation (see Klin & Jones)
- Toddlers with clear-cut symptoms at 18 months tend to have stable diagnoses. Particularly predictive:
  - Poor e.c. + limited gestures + limited giving/showing
  - Okay e.c. + repetitive behavior + limited giving/showing
- Differences in attending to social stimuli & sticky attention, even very early on
- Infant siblings are also at risk for developmental problems, along with "broader autism phenotype" presentations
- Infant siblings are responsive to early intervention (Schwichtenberg et al, 2018; Green et al, 2015; Green et al, 2017)

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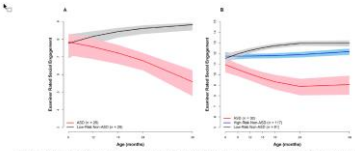
Infant Sibling Studies



- "Small disruptions of early social adaptive action can yield progressively greater between-group differences over time, culminating in the marked symptomatology of ASD." (Schultz et. al, 2018)
- Understanding the social communication trajectory is important to intervening earlier
- How can we use this in surveillance/screening?
  - Observation (e.g., Social Communication growth curves)
  - Parent surveillance (e.g., Learn the Signs. Act Early.)

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Traditional Autism Diagnostics

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Diagnostic interviewing

- Families are thoroughly interviewed about the child's communication and social deficits as well as the child's restricted interests and repetitive behaviors.
- Both present and past concerns are considered.
- The Autism Diagnostic Interview, Revised (ADI-R) is considered the "gold standard" for structured diagnostic interviewing.
- Sometimes daycare providers, preschool teachers, or other adults who know the child well are also briefly interviewed.

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Structured observation

If there are concerns, most children complete a semi-structured observation.

The "gold standard" measure is the Autism Diagnostic Observation Schedule, Second Edition (ADOS-2).

- ADOS-2 activities elicit behaviors pertinent to the diagnosis (e.g., eye contact, play and imaginative skills, initiation of social games).
- Behavioral observations are then scored according to specific guidelines.
- ADOS-2 activities vary by the child's age and language abilities.

There are alternate measures that can be used (e.g., Tele-ASD-Peds, CARS-2), but the ADOS-2 is very popular.

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ADOS-2 Toddler Module

- Can be reliability used with infants/toddlers with nonverbal cognitive skills ~12 months to age 30 months.
- ADOS Toddler Module

Social
- Limited responsive social smiling
- Limited response to name
- Limited initiation of and response to joint referencing
- Poorly coordinated eye gaze and/or limited eye gaze
- Limited directed facial expressions
- Limited interest/shared enjoyment in non-physical social games
- Limited showing/giving

Communication
- Limited communication bids, including gestures
- Atypical vocal intonation
- Echolalia

Behaviors & Interests
- Unusual sensory interests
- Repetitive mannerisms
- Nonfunctional & repetitive play, with limited interest in imitating others' play

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Estimates of developmental functioning:

- Will frequently need to review or obtain cognitive, language, and other developmental evaluations (e.g., adaptive functioning, motor functioning) in order to contextualize the social communication deficits demonstrated by the child.
- These estimates can also be important for intervention planning as well as for making a differential diagnosis.

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What is the future of autism diagnostics?

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Using fewer/different tools

Not every patient suspected of autism needs the gold standard measures.

There is no requirement in the official diagnostic criteria that testing must be done in order to diagnose.

Insurance companies keep patients with autism out of needed intervention services by stating they need certain tests or certain scores in order for their diagnosis to count (this is illegal).

"….best practice processes for diagnostic assessment exist independently of the use of any single tool. Standardized diagnostic instruments were developed to aid the expert, to structure the assessment to ensure that diagnostically relevant information would be available for making diagnostic judgments. Never were they meant to prevent access to appropriate services" (Bishop & Lord, 2023)

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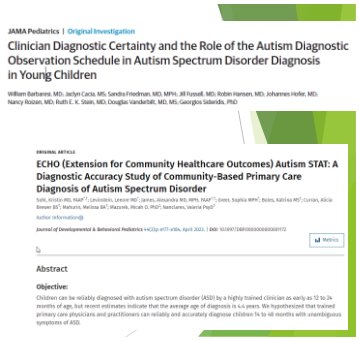
Using fewer specialists

- DBPs' impressions matched the ADOS evaluation 90% of the time.
- "Diagnostic certainty" predicted match
- In Missouri, a project trained primary care providers to diagnose frank autism accurately using the STAT.
  - who educates and follows up?



JAMA Pediatrics | Original Investigation

Clinician Diagnostic Certainty and the Role of the Autism Diagnostic Observation Schedule in Autism Spectrum Disorder Diagnosis in Young Children

William Barbaresi, MD; Jaclyn Cacia, MS; Sandra Friedman, MD, MPH; Jill Fussell, MD; Robin Hansen, MD; Johannes Hofer, MD; Nancy Nixon, MD; Ruth E. K. Stein, MD; Douglas VanderBelt, MD, MS; Georgios Sideridis, PhD

ORIGINAL ARTICLE

ECHO (Extension for Community Healthcare Outcomes) Autism STAT: A Diagnostic Accuracy Study of Community-Based Primary Care Diagnosis of Autism Spectrum Disorder

Abstract

Objective:

Children can be reliably diagnosed with autism spectrum disorder (ASD) by a highly trained clinician as early as 12 to 24 months of age, but recent estimates indicate that the average age of diagnosis is 4.4 years. We hypothesized that trained primary care physicians and practitioners can reliably and accurately diagnose children 14 to 48 months with unambiguous symptoms of ASD.

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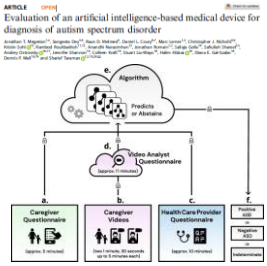
Using AI



ARTICLE OPEN

Evaluation of an artificial intelligence-based medical device for diagnosis of autism spectrum disorder

CANVAS-DX DETERMINANT (32%)

- Sensitivity = 98.4%
- Specificity = 78.9%

- If you included the whole sample, the metrics are different:
  - Sensitivity = 51.6%
  - Specificity = 18.5%

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Autism Interventions

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Interventions are done with goals in mind

- Individualized
  - Improve communication, social functioning, and play skills
  - Improve adaptive functioning
  - Promote academic functioning
  - Decrease challenging behaviors
  - Address comorbid medical and psychiatric conditions
  - Promote independence and quality of life
- Written collaboratively & with progress assessed/shared routinely
- Short-term treatment goals provide scaffolding to long-term outcomes

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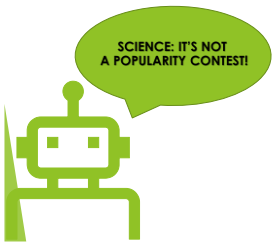
Why Evidence-Based?



- Overwhelming number of interventions for autism.
- Some interventions have little research showing that they work.
- Time and money are limited.

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Sources of Consensus Regarding ASD Treatment Effectiveness

- American Academy of Pediatrics (AAP) Council on Children with Disabilities
  - https://pediatrics.aappublications.org/content/145/1/e20193447 (revised guidelines published in 2020)
- National Autism Center – National Standards Project
  - http://www.nationalautismcenter.org/national-standards-project/ (phase 2 was completed in 2015, phase 3 overdue)
- Association for Science in Autism Treatment
  - http://www.asatonline.org/
- National Clearinghouse on Autism Evidence and Practice
  - https://ncaep.fpg.unc.edu/ (new summary published in 2020)

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Interventions Supported by Scientific Evidence

- Applied Behavior Analysis (ABA), including Discrete Trial Training and Functional Communication Training (FCT)
- Naturalistic Developmental Behavioral Interventions (e.g., Project ImPACT, Early Start Denver Model)
- Cognitive Behavior Therapy (CBT) and Self-Management
- Social skills training, including peer-based strategies, social stories, scripting, and social skills groups
- Language training

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ADDITIONAL STRATEGIES Supported by Scientific Evidence

- Visual supports and schedules
- Naturalistic teaching strategies
- Modeling, live or video
- Parent training

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Promising or Emerging Treatments

- Treatment and Education of Autistic and related Communication-Handicapped Children (TEACCH)
- Technology-based treatments, using computers or other electronic devices
- Music therapy (close to fully established)
- Developmental relationship-based therapies, such as Floortime (PLAY Project)

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Treatments with Limited Scientific Evidence

- Gluten and casein-free diets
- Facilitated communication
- Auditory integration training

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I recommend and use interventions that I think:

(1) are most likely to achieve short-term goals and work toward desired outcomes,

(2) are available, &

(3) are a good fit for the patient/family.

With luck, there are multiple options available to the patient/family without having to travel or move.

http://interactingwithautism.com/video/rogers_0014

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