UCDAVIS HEALTH | MIND INSTITUTE

Identification of Autism in Primary Care Pediatrics with Discussion about Capacity-Building at the Local Level

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Disclosures

- I have no financial relationship with any manufacturer of any commercial product and/or provider of commercial services discussed in this CME activity
- I do not intend to discuss an unapproved/investigative use of a commercial product or device in our presentation
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Overview

Overview of Autism Spectrum Disorder

- Epidemiology of ASD. Disparities related to access to evaluation
- Typical social communication and play in young children
- Description of early ASD symptoms in young children
- Overview of Developmental Screening and Screening for ASD

Review of Diagnostic Assessment and Expanding Role of Medical Home

- How we diagnose ASD
- How to prepare your patients for an ASD evaluation- it's play based
- How long it takes, "paperwork" burden, multiple visits, access to care
- Could we diagnose more children within the Medical Home?
  - Evolving Models

Resources, Referrals, Schools

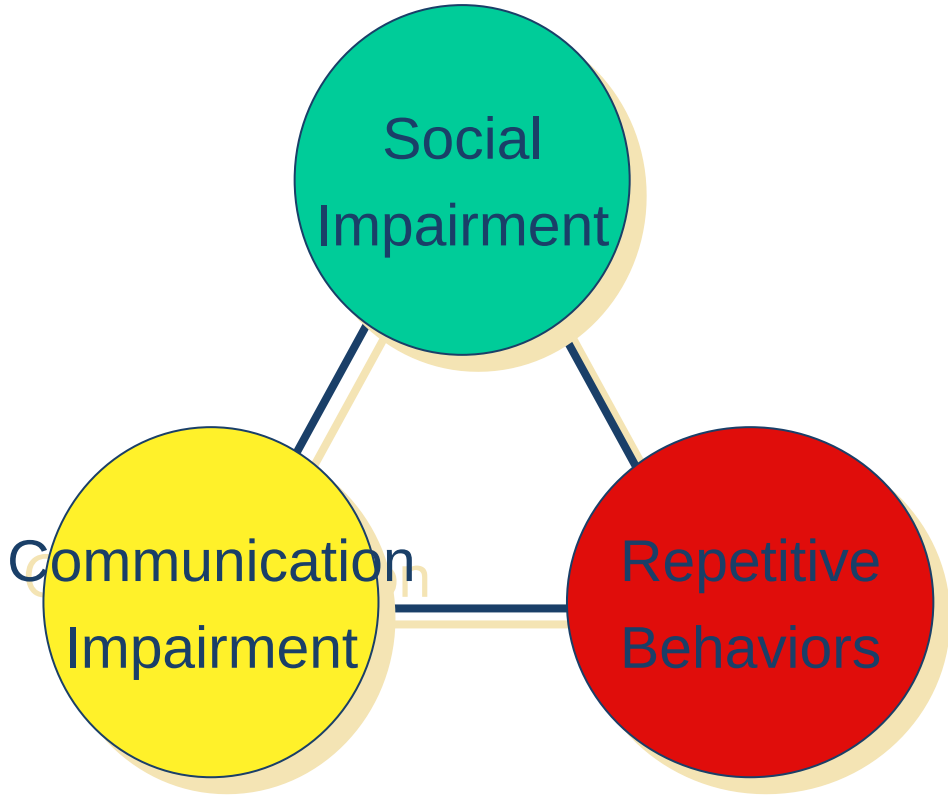
- Role of Schools
- Role of California Regional Center and Early Start Program
- Community Based Agencies, Parent Support Groups

What is Autism?

- A group of disorders with:
  - Common core behavioral symptoms
  - Wide variability of symptoms
  - Multiple etiologies
  - Neurologic basis
  - Syndromes
- ***NOT: Vaccines, food allergies, metal intoxication, fungal infection, parenting factors***

Autism Core Features

- **Severe and pervasive** impairments in 3 areas of development
  - **Reciprocal social interactions**
  - **Verbal and nonverbal communication**
  - **Presence of stereotyped behaviors, restricted interests, sensory dysregulation**

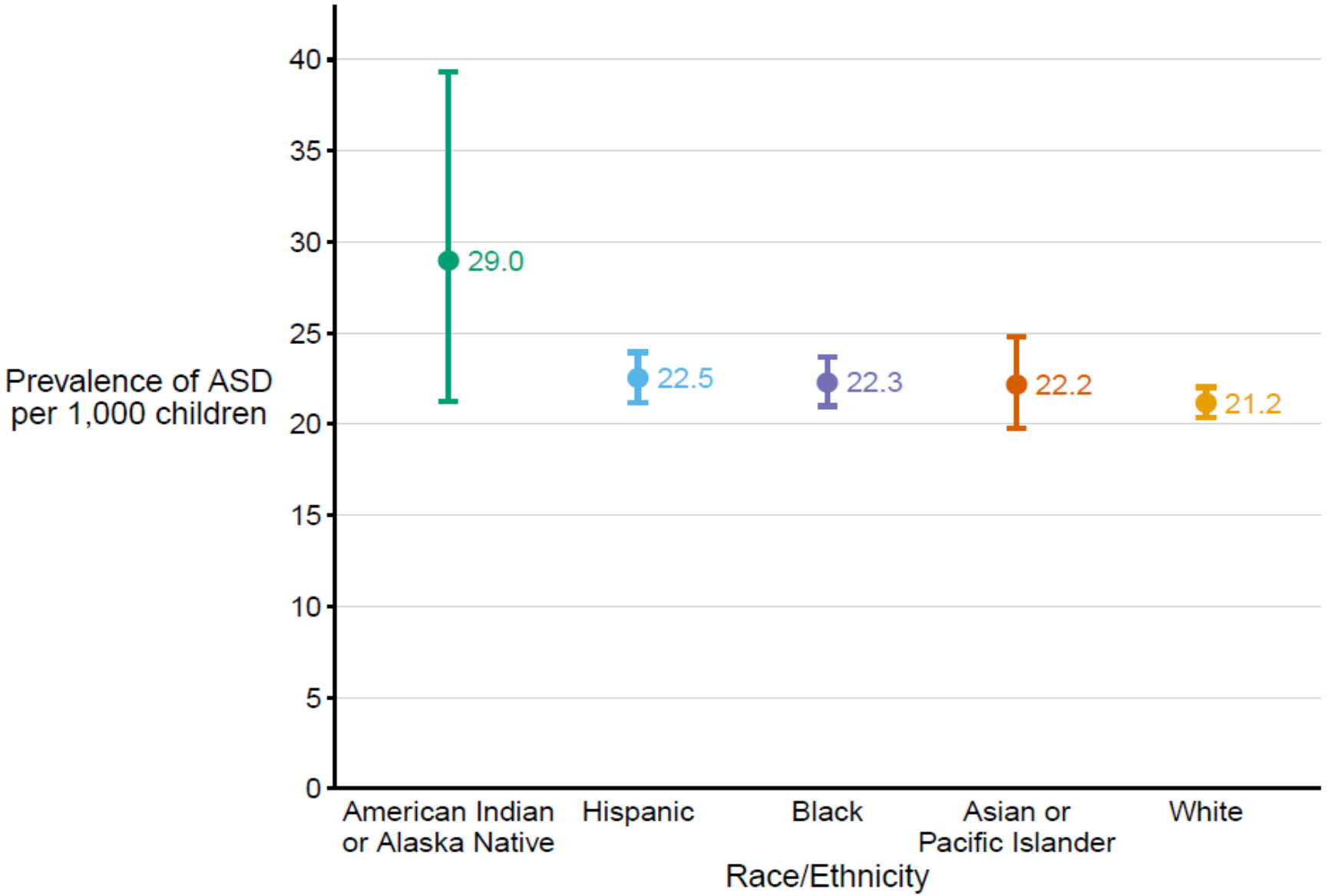


Epidemiology
Autism Spectrum Disorders

- Prevalence increasing worldwide
  - Asia, Europe, and North America average prevalence of between 1% and 2%
- Prevalence of ASD in 8 year olds in USA
  - 1 in 150 US children (CDC, 2002)
  - 1 in 110 US children (CDC, 2006)
  - 1 in 88 US children (CDC, 2008)
  - 1 in 54 US children (CDC, 2016)
  - 1 in 44 US children (CDC, 2018)
  - 1 IN 36 US children (CDC, 2020)

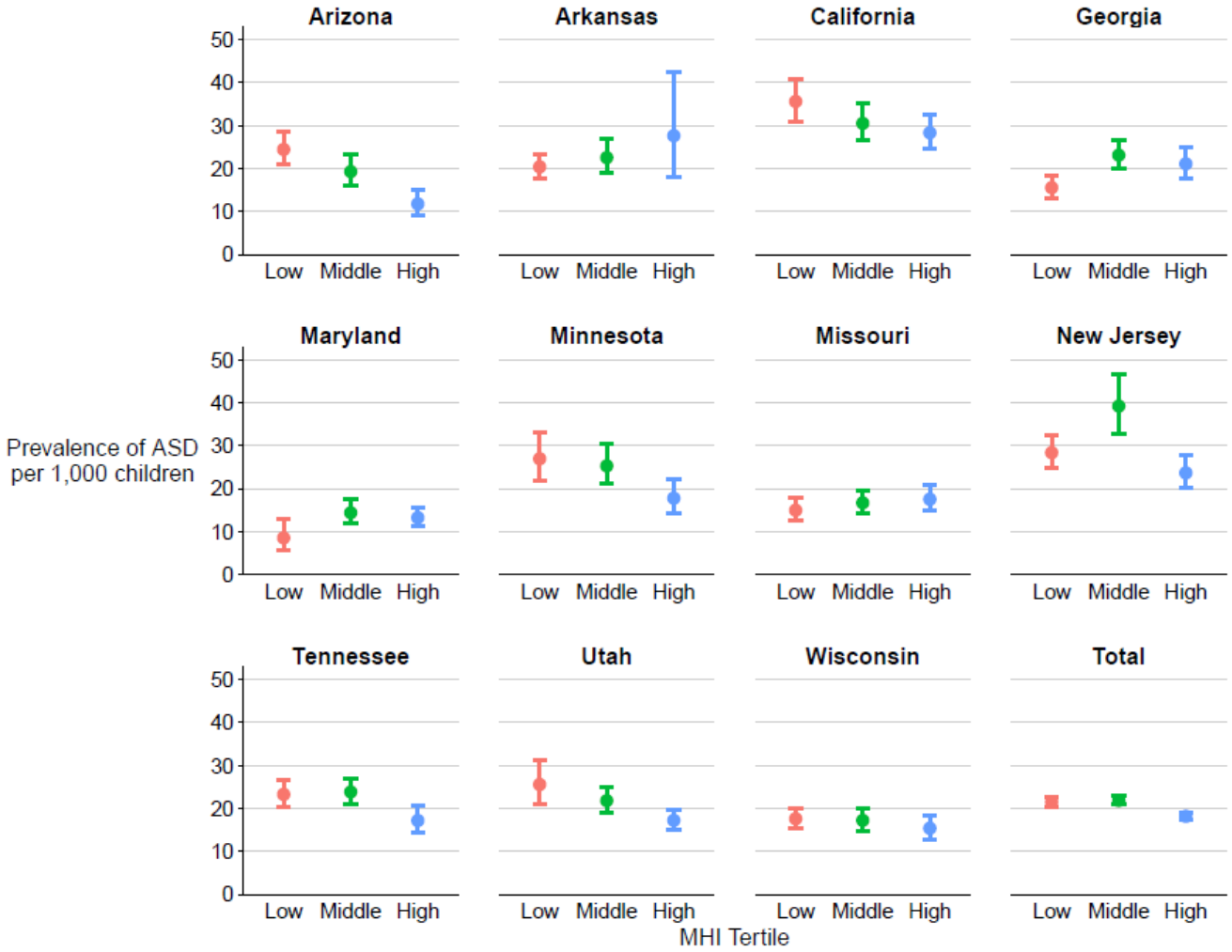
Prevalence of autism spectrum disorder per 1,000 children aged 8 years, by race/ethnicity

Autism and Developmental Disabilities Monitoring Network, 11 sites, United States, 2018



Prevalence* of autism spectrum disorder per 1,000 children aged 8 years, by median household income tertile and site†

Autism and Developmental Disabilities Monitoring Network, 11 sites, United States, 2018

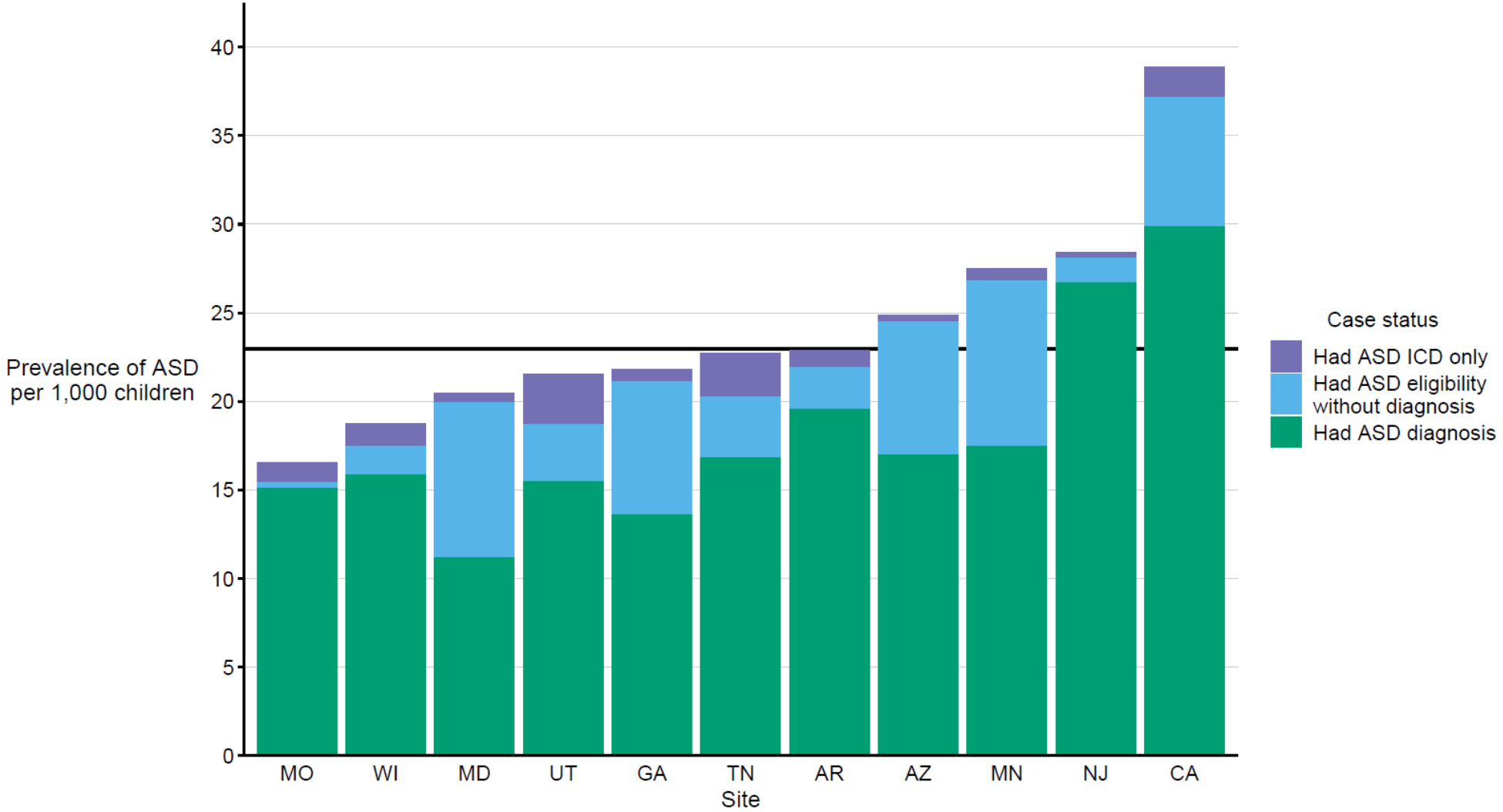


* Dots are the point estimates and horizontal lines are the 95% confidence intervals.

† Cochran Armitage test of trend results for association between socioeconomic status tertile and ASD prevalence, by site and overall: Arizona (p<0.001), Arkansas (p = 0.17), California (p = 0.03), Georgia (p = 0.01), Maryland (p = 0.21), Minnesota (p = 0.01), Missouri (p = 0.21), New Jersey (p = 0.15), Tennessee (p = 0.02), Utah (p<0.001), and Wisconsin (p = 0.27); all sites (p<0.001).

Prevalence* of autism spectrum disorder per 1,000 children aged 8 years, by identification type and site

Autism and Developmental Disabilities Monitoring Network, 11 sites, United States, 2018



* Horizontal line is the overall Autism and Developmental Disabilities Monitoring Network prevalence of 23.0 per 1,000 children aged 8 years. Children with documented ASD statements could also have ASD classifications in special education or ASD ICD codes.

Variable Access to Assessment, Significant Disparities

- The average age of ASD diagnosis in African American children was 64.9 months (±49.6), on average 42.3 months (±45.1) after parents' first concerns about their children's development
- Although similar rates of ASD prevalence are reported in both rural and urban areas, individuals in rural communities, report limited access to resources and timely identification and intervention services
- About 1 in 6 (17%) US children aged 3–17 years were diagnosed with a developmental disability (2017)

Constantino, et al, 2020, Mohamed et al., 2016 , Cohen and Hesselbart, 1993; Slade, 2003; Green et al., 2013; Gona et al., 2016

What causes Autism Spectrum Disorders?

- Almost half (approx. 45%) of children identified with ASD have average to above average intellectual ability (IQ)
- ASD is about 4 times more common in boys than girls
  - Maybe…. long history of underdiagnosis in girls
  - Presentation is different
- Children born to older parents are at a higher risk for ASD
- ASD commonly co-occurs with other developmental, psychiatric, neurologic, chromosomal, and genetic diagnoses.
- ASD are neurodevelopmental conditions with a genetic component
  - Parents who have a child with ASD have a 10 to 20% chance of having a second child with ASD

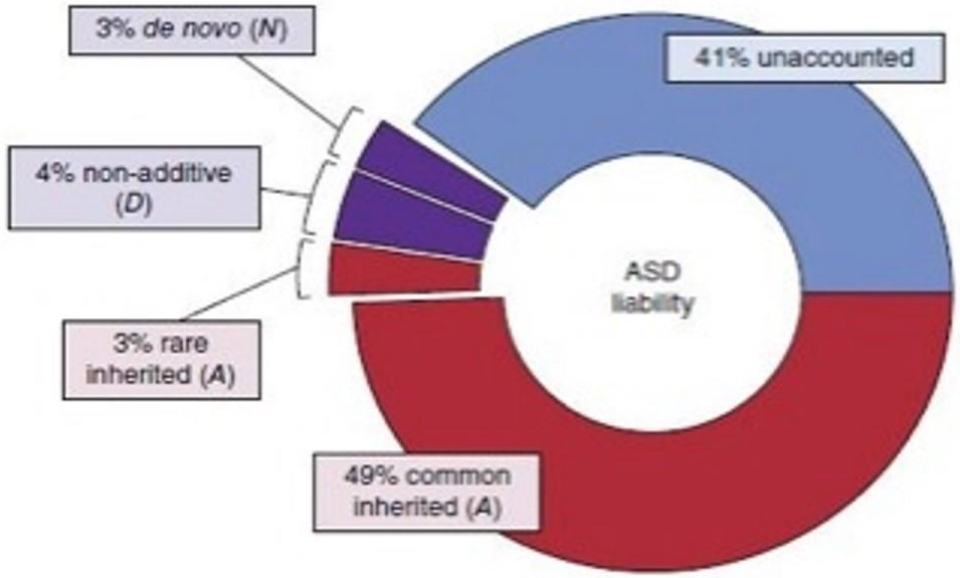
(CDC May 2018)

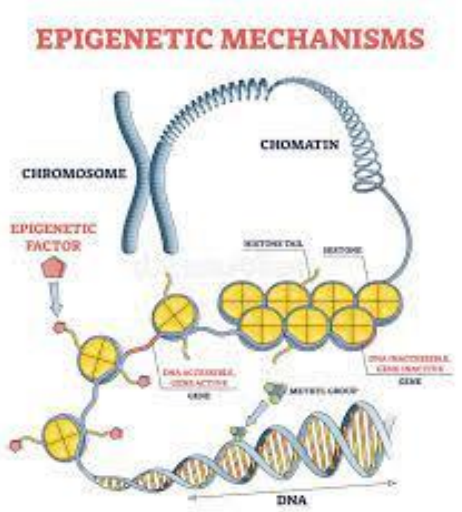
Landmark Twin Study (Risch, et al., 2011)

- 192 pairs of twins
  - 54 were identical (genetically IDENTICAL)
  - 138 fraternal (fraternal twins are genetic siblings)
  - The initial results changed our thinking about etiology
    - 31% of male ***fraternal*** twins both have ASD
    - 36% of female ***fraternal*** twins both have an ASD
    - 77 % of male ***identical*** twins both have an ASD
    - 50 % of female ***identical*** twins both have an ASD
  - Environment represented more than half of susceptibility (55-58%)
  - Genetics represented 38% represented of the risk

Inherited Risk is Seldom the Whole Picture

- A few inherited genes are sufficient by themselves to cause autism (about 6%)
- But most "autism genes" only increase the risk that an individual will go on to develop ASD
- Autism most often results from a combination of genetic susceptibility and environmental triggers





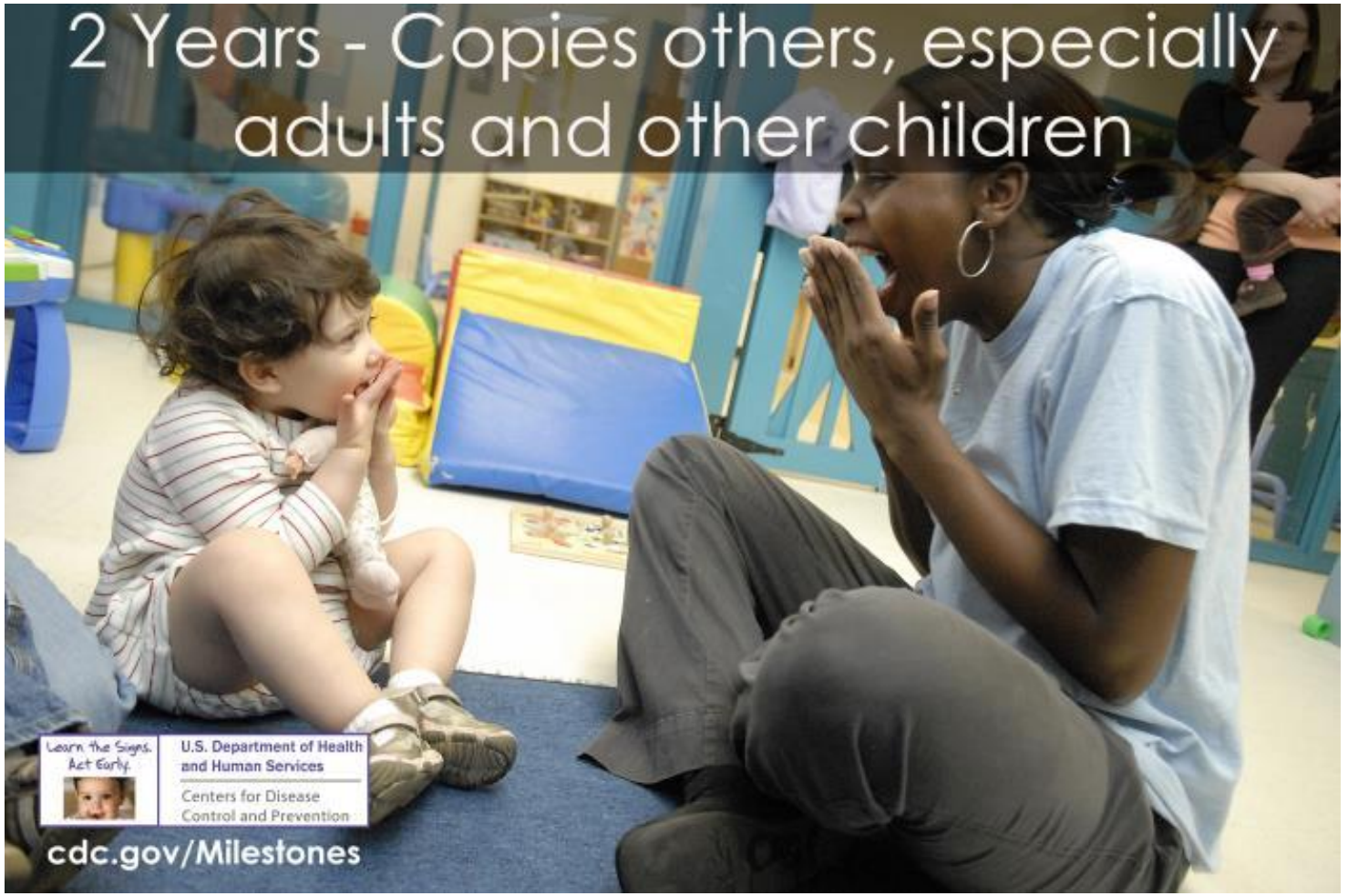
Typical Development

- By **18 months of age**, children typically
  - Respond to their name
  - Point to request and show
  - Give and show objects to others
  - Look when a caregiver points or turns their head to look at something
  - Share affect, show enjoyment in language-gesture routines
  - Imitation- copy caregiver's actions
  - Say about 5 words, including mama and dada
  - Functional Play (phone, hairbrush, cup, ball)

Typical Development

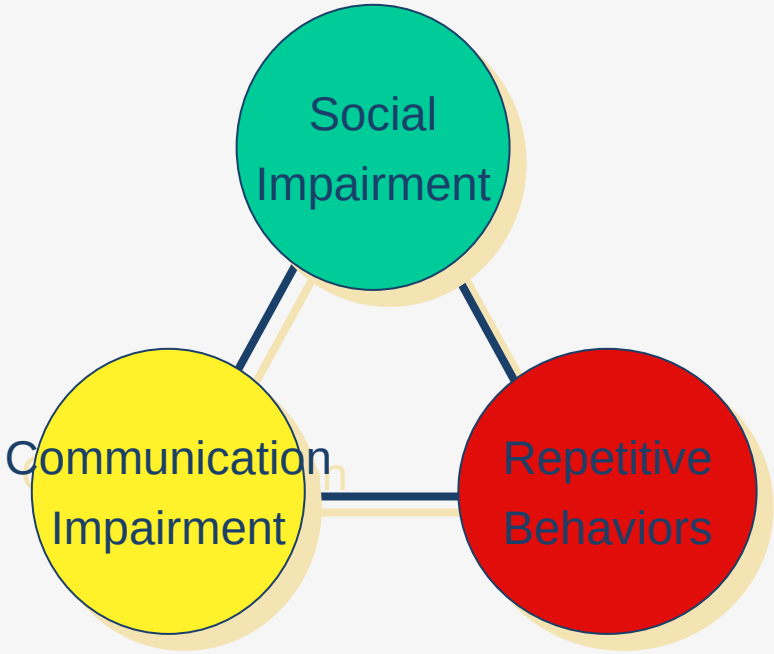
Give, show and point to request by 18 months

Typical 18-month communication- Few words, but communicative intent is clear!


2 Years - Copies others, especially adults and other children
Learn the Signs. Act Early.
U.S. Department of Health and Human Services
Centers for Disease Control and Prevention
cdc.gov/Milestones

What does ASD look like in Younger Children

- Let's think about 18, 24 and 30 month old month well child visits
  - MCHAT-R
  - Ages and Stages, SWYC (Survey of Well Being in Young Children) or other screening tool

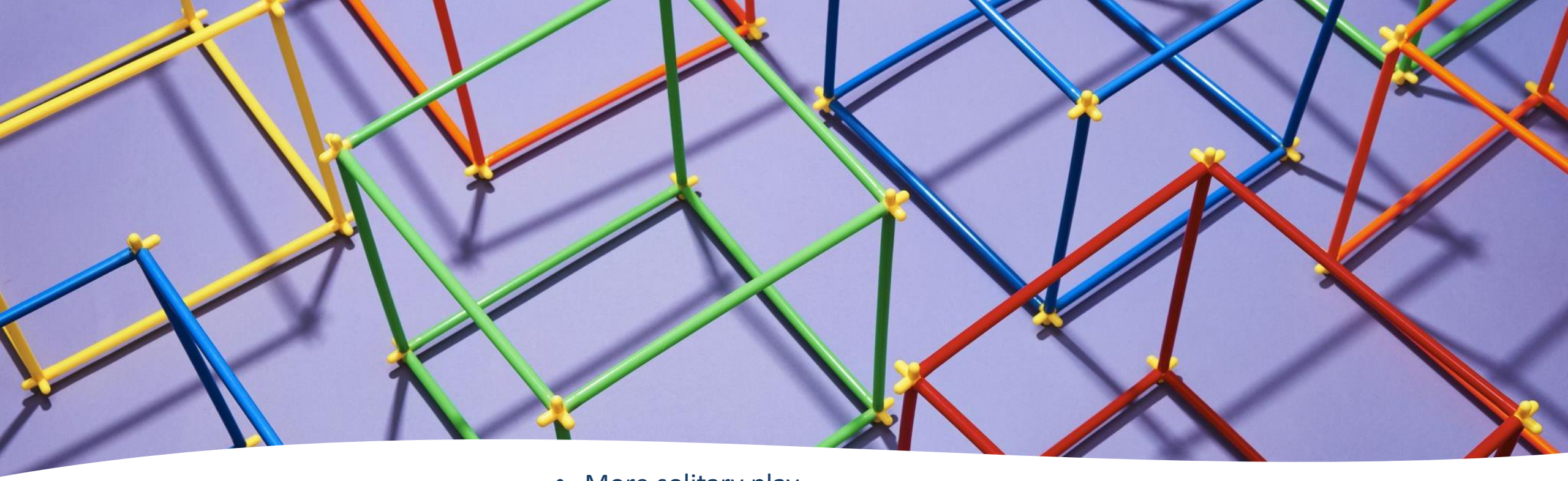


Communication in Children with ASD

- Limited communicative intent
  - Remember the previous examples of clear communicative intent
  - Vocalizing without requesting-language that lacks function
- Atypical vocalizations, echolalia
- Decreased use of gestures
  - Less likely to wave bye bye, high 5, nod or use descriptive gestures
- Hand leading, using another's hand as a tool
  - Blocking task
- Language delay
  - Few words or only words used repetitively, no signs
- Less likely to be interested in language gesture games
  - Peek a boo, wheels on the bus, the dreaded Baby Shark

Social Interaction in Children with concern for ASD

- Limited response to name
- Fewer attempts to draw others attention, share the moment
  - Less frequent pointing, showing, giving
- Decreased response to joint attention- follows a point or gaze shift
- Eye contact that is not well modulated with vocalization, gestures and facial expression



Play in ASD

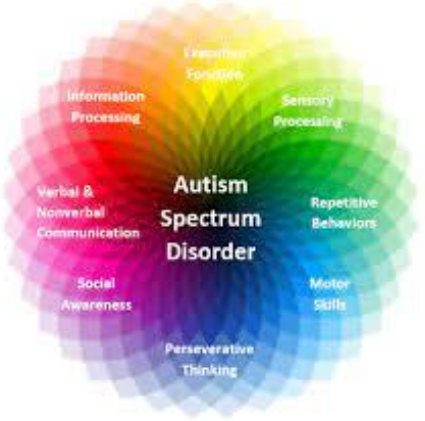
- More solitary play.
  - Often “self-contained” will play alone for prolonged periods of time
- Less imitation
- Less functional or pretend play
- Developmentally inappropriate play
  - Sensory input>>cause and effect>>functional>>pretend>>collaborative
- Lining up, arranging, sorting, carrying and prolonged inspection of objects

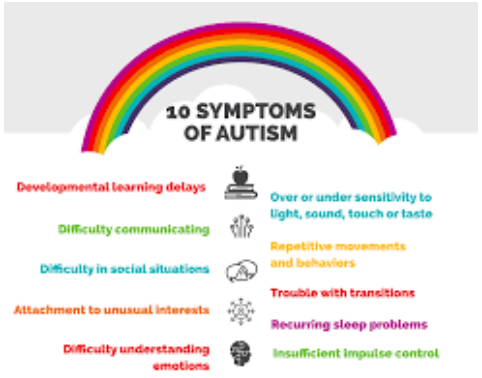
Repetitive Behaviors/Restricted Interests

- Motor Stereotypies
- Repetitive play, atypical play
- Sensory seeking or sensory dysregulation
- Behavioral rigidity, difficulty accepting change, insistence on routine
- Restricted, fixated interests

Videos courtesy of Autism Navigator

- Comparison of Social Emotional Reciprocity
  https://resources.autismnavigator.com/asdglossary/#/section/9/initiateInteraction
- Repetitive, stereotypic motor movements
  https://resources.autismnavigator.com/asdglossary/#/section/21/repetitiveMotor
- Fixated interests, non-functional play, perseveration
  https://resources.autismnavigator.com/asdglossary/#/section/28/unusualObjects





How do we decide symptom severity and threshold for diagnosis?

DSM-V Criteria ASD

- A. Persistent deficits in social communication and social interaction across multiple contexts. Must meet criteria for all 3 areas
  - Social-emotional reciprocity (connectedness)
  - Nonverbal communication behaviors used for social interaction (gestures, eye contact, facial expression)
  - Developing, maintaining, and understanding relationships (friends, sibs, caregivers)

- Providers assign a level of severity
  - Level 3-Requiring very substantial support
  - Level 2- Requiring substantial support
  - Level - Requiring support

DSM-V Criteria ASD

B. Restricted, repetitive patterns of behavior, interests or behavior as manifested by at least 2 of 4 of the following:

- Stereotyped or repetitive motor movements, use of objects, or speech
- Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or non-verbal behavior
- Highly restricted, fixated interests that are abnormal in intensity or focus
- Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment

C. Present in early development

D. Causes significant impairment

E. Not better explained by Intellectual Disability, Global Developmental Delay

DSM-5 Diagnostic Criteria

- Now just Autism Spectrum Disorder (ASD)

  Eliminated previous sub-categories

  - Autistic Disorder
  - Pervasive Developmental Disorder – Not Otherwise Specified (PDD-NOS or PDD)
  - Asperger Syndrome
  - Childhood Disintegrative Disorder (CDD)

  These conditions are now lumped together under ASD

  Many Neurogenetic Disorders used to be "CDD"

  - Rett Syndrome: example of a neurodegenerative disorder better described as a distinct neurogenetic disorder

Typical Child referred to us at 18-36 months of age

- 24-month-old well child visit….with you!
  - Parents very concerned about development. At his/her 2-year-old birthday party, he would not join other kids in play, was the only child not yet speaking, cried when the other children sang “Happy Birthday” and refused to blow out candles, “all the other kids were able to sing along.” My friend is a teacher and she thinks he/her might have autism.
- Brief history
  - Not yet using words to communicate but will repeat words.
  - When he wants something, he just gets it himself or pulls parents by the hand to where it is and cries.
  - “Plays on his own for hours.”
  - Hates loud noises, very upset with vacuum/toilet flushing/blender and washing face, brushing teeth
  - Will “flap hands” when excited or upset.
  - Play- he likes to organize toys, groups them and likes to look at them closely, likes to take objects in/out of containers, likes some puzzles, loves songs and shows about numbers on iPad. No functional play or pretend play
- Physical Exam, Medical History, Birth History unremarkable
- ROS
  - Some difficulty falling and staying asleep, picky eater, but growth is ok and you think he has adequate sources or iron, calcium and protein in his diet.

Early Identification is Critical

Early signs of ASD are usually present by 18-24 months.

- Screening tools recommended (MCHAT-R, SWYC, Ages & Stages, PEDS: DM)

Early intervention works

Evidence based treatments for ASD are widely available

Do NOT take a "wait & see" approach

- AAP CPG recommends:
  - Ongoing developmental surveillance
  - Formal developmental screening at 9, 18 and 24 or 30 month well child visits and autism-specific screening at 18 and 24 or 30 months
- If concerns or failed screen, refer for developmental evaluation/early intervention
  - Autism Diagnostic Observation Schedule 2nd ed.
  - Developmental/cognitive testing
- Audiology examination
- Speech/occupational/physical therapy referrals if indicated

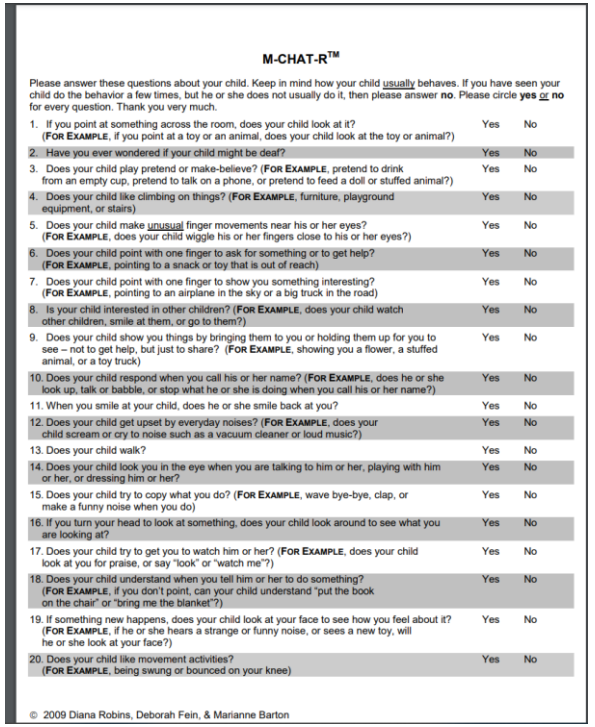
Source: AAP Council on Children with Disabilities et al. *Pediatrics*, 2006

Screening tools

Measure	Ages	Items	Cost
Modified Checklist for Autism in Toddlers-R/F (M-CHAT-R/F)	16-30 months	20 items, Y/N, parent completed tiers: low risk (0-2), medium risk (3-7), high risk (8-20)	Free
The Survey of Well-Being of Young Children (SWYC): Parent's Observations of Social Interactions (POSI)	16-35 months	6 items parent completed Integrated into SWYC	Free
Screening Tool for Autism in Toddlers and Young Children (STAT)	24-35 months	12 items provider completed Interactive/observational	See web
Rapid Interactive Screening Test for Autism in Toddlers (RITA-T)	18-36 months	9-item provider-completed Interactive/observational	See web
Communication and Symbolic Behavior Scales Developmental Profile – Infant/Toddler Checklist (CSBS-ITC)	6 to 24 months	24-item multiple choice parent checklist	Free
Social Communication Questionnaire (SCQ)	4+ years	40-item parent completed checklist; must have a mental age of at least 2 years	See web

M-CHAT-R

- 16 to 48 months of age
- Available in many, many languages
  - https://mchatscreen.com/mchat-rf/translations/
- Free download
  - https://www.autismspeaks.org/screen-your-child
- 5 min to screen, 5 min to score
- Questionnaire completed by parent: 23 yes/no items



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 unusual 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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Typical Child referred to us at 18-36 months of age

- 24-month-old well child visit
- At 18 months, MCHAT was 5 (0-2 low risk, **3-7 medium risk**, >8 high risk)
  - Not responding to name, not following a point, not playing pretend of make believe, makes unusual finger movements near his/her face, not pointing, not interested in other children
- Ages and Stages with delays in **communication and personal social domains**
  - Not yet using words to communicate, but will repeat words
    - When he wants something, he just gets it himself or pulls us to where it is and cries.
    - "Plays on his own for hours"
    - Very upset with vacuum/toilet flushing/blender and washing face, brushing teeth, will "flap hands" when excited or upset.
    - Play- he likes to organize toys, groups them and likes to look at them closely, likes to take objects in/out of containers, likes some puzzles, loves songs and shows about numbers on ipad.
    - No functional play or pretend play
- Seen by a very well-trained PCP or EI provider and referred to Early Start through Regional Center AND developmental pediatrics or child psychology for ASD assessment

ASD evaluations use common toys and familiar play

- Response to name
- “Peek a boo” or “I’m gonna get you”
- Pop up toy, shape sorter, blocks
- Bubbles
- Balloon or rockets

Traditional ASD Evaluation in Children <5

- Medical History
- Developmental History
  - Intake form
- Assessment Questionnaires
  - Adaptive function
  - Developmental Level
  - Behavior rating scales
  - Autism Symptom Checklists
- ASD specific assessment and cognitive or developmental testing
  - Autism Diagnostic Observation Schedule (ADOS-2)
  - DSM V Based assessment with behavioral observations
  - 2-4 hours of evaluation time!
  - Driving distance, time off work, access to care limited, long wait lists

- Behavioral treatments: remain the backbone of treatment
  - Applied Behavior Analysis
  - Other similar treatments (ESDM, Floortime, TEACCH)
- Speech therapy, occupational therapy, social skills groups
- Parent support groups
- Could be more than 25 hours per week of therapies
  - Think about anything you do for more than 25 hours per week that you don't get paid to do…it's a lot to ask

Alta California
Regional Center

Early Intervention Services

Assisting Families and Children

Early Intervention is not a cure for disabilities or risk conditions. Rather, it is a system of coordinated services that promotes the child's growth and development and supports families during the critical early years.

Early Intervention Services are:

- Are designed to meet the developmental needs of each eligible child and the needs of the family related to enhancing the child's development;
- Are selected in collaboration with the parents;

Medial Evaluation

- Hearing Screening-all kids with significant language delay
- Physical exam
- Chromosomal Microarray and DNA sequencing for fragile X syndrome
  - Genetic causes 10-30%-can offer Tier 1 evaluation after diagnosis of ASD
- Consider co-occurring conditions seizures, dysmorphology, sleep, GI
- Screen for anxiety, ADHD, learning disabilities, intellectual disability
  - >95% 4–8-year-olds have at least 1 co-occurring issues
  - Swedish study- 50% of children with ASD had >4 co-occurring conditions

Common Medical Concerns

- Self-restricted diet-can be secondary to sensory issues and behavioral rigidity or underlying GI pathology (EoE, GERD, hiatal hernia, IBD, celiac)
- Behavioral insomnia-should rule out organic sleep issues
- Elimination
- Hypotonia
- Pica
- Seizures
- Medical issues presenting as change in behavior (esp in non-verbal patients)
- Sensory issues may impact pill-swallowing
- Very high rates of medical trauma>>> care refusal>>>unmet healthcare needs

Our patient who was referred at 24-month-old well child visit

- Referred for audiology (normal) at 26 months of age and to Regional Center for Speech Therapy
- Began speech therapy and support from early childhood educator at 27 months of age
  - Therapists are working on imitation, joint attention, play skills
  - He is now using signs for “more” and “all done” and he has about 5 words. He is starting to imitate therapists and his caregivers in games like itsy bitsy spider and smiles and laughs during these games and loves Baby Shark
- Evaluated by Developmental Pediatrician at MIND Institute at 28 months of age
- Met DSM-V criteria for ASD
- Referrals placed to ABA therapy, a Family Navigator is assigned to caregivers, parents begin an 8-week caregiver coaching program with basics of ASD
  - He starts ABA at 30 months of age (6 months later, if everything went well)
- Sleep, Feeding Issues begin to be addressed in coordination with PCM
- Chromosomal microarray and fragile X testing done and are non-diagnostic

Questions?

Discussion about evolving models that expand capacity to evaluate young children for concerns about ASD

Early Intervention Supports vs. School Based “Special Education”

0-3 years- Services through Part C Federal Funding

- Free-Federally Funded in all states
  - *In CA Early Start via Regional Center*
- Family centered, in home, coaching model
- NOT MEDICAL
- Goal: minimize impact of disabilities on the child’s overall development

3+ years-Services in public school system

- Free-must be offered in least restrictive environment
- Therapies like speech therapy or occupation therapy are provided through school district
- Children with autism also can almost always participate in a preschool program with specialized instruction informed by ABA.
- Child centered
- Focuses on education

ASD Services Covered by Health Insurance

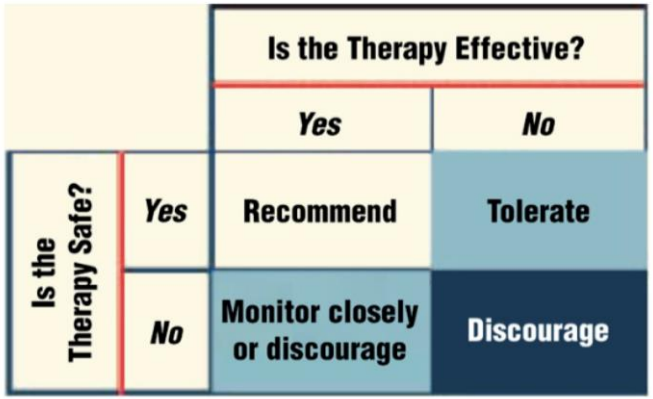
- Specialty Medical Assessments
- ABA
- Genetic Testing
- Speech Therapy for medical conditions (apraxia, cleft palate, feeding disorder, VPI)
- Physical Therapy or Occupational Therapy for conditions that are not educationally related (needs DME or orthotics or OT for help learning to dress or toilet, but hand writing ok)

Psychopharmacology

- Good evidence for risperidone and abilify in ASD, but only for aggressive behavior, self-injury, property destruction
  - Significant side effects
    - Obesity, hyperphagia, metabolic syndrome with diabetes, gynecomastia
- Medications may be used to target symptoms, but with limited evidence of efficacy in ASD
  - SSRI's
  - Stimulants (limited evidence of efficacy in ASD)
  - Alpha-adrenergics
  - Sleep Aids

Complementary and Alternative

- Encourage open dialogue and disclosure of CAM use with healthcare providers
- Melatonin is effective for helping kids fall asleep
- No evidence for:
  - High dose vitamins or probiotics
  - GFCF or other diets
  - Chelation Therapy
  - Immunologic Therapy
- Support families in gathering evidence based information
- Families who delay vaccination need support
- Direct them to reliable sources of information
- National Center for Complementary and Integrative Health
  - https://nccih.nih.gov/
- U.S. National Library of Medicine
  - https://medlineplus.gov/complementaryandintegrativemedicine.html



Kemper KJ, Cohen M. Ethics meet complementary and alternative medicine: new light on old principles. *Contemp Pediatr*. 2004;21(3):61

Local Autism Resources
Online Modules for Families

MIND Institute Center for Excellence in Developmental Disabilities

Support groups, sibling workshops, teacher training, advocacy, Tip Videos

https://health.ucdavis.edu/mindinstitute/centers/cedd.html

Autism Speaks Tool Kits

Haircuts, dental visit, challenging behaviors, toilet training

https://www.autismspeaks.org/tool-kit?resource_type[606]=606&resource_type[606]=606&state[186]=186

Help Is in Your Hands

Resource for parents and providers who work with children with concerns about delays in social communication, immature play skills and repetitive behaviors in a naturalistic way to help children learn to attend to others and build play and communication skills. The modules can be accessed at:

helpisinyourhands.org

Autism Distance Education Parent Training (ADEPT Modules)

Online ADEPT modules are accessible and free to families and providers to help parents of children with autism with regards to behavioral and self-help skills:

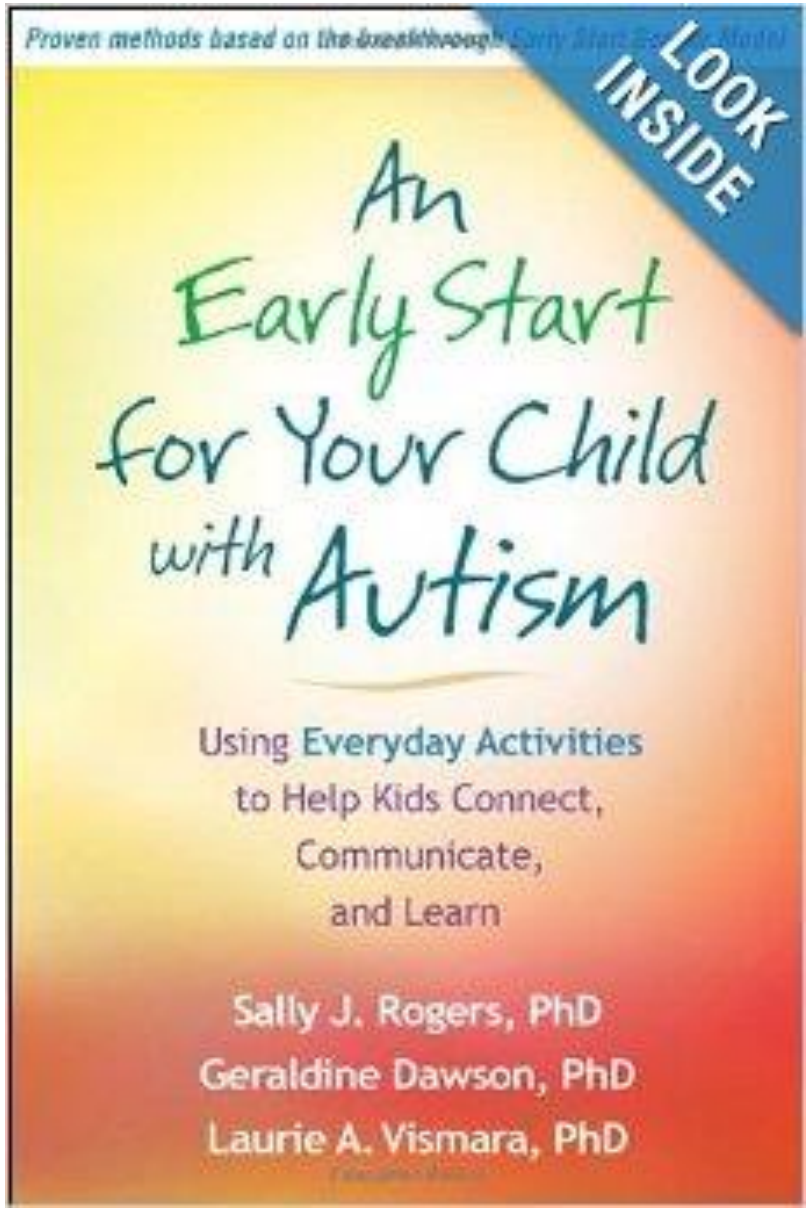
https://health.ucdavis.edu/mindinstitute/centers/cedd/adept.html

Resources for Children with NDD

- Early Start Program in California (0-36 months)
  - http://www.dds.ca.gov/EarlyStart/ESQuestionAnswers.cfm
  - 800-515-BABY
- California Regional Centers
  - http://www.dds.ca.gov/rc/rclist.cfm
  - 0-3 Early Intervention Program
- California Children Services
  - http://www.dhcs.ca.gov/services/ccs/Pages/ProgramOverview.aspx
- CDC Learn the Signs Act Early
  - https://www.cdc.gov/ncbddd/actearly/index.html

Resources

- Autism Speaks First 100 days toolkit
  - https://www.autismspeaks.org/family-services/tool-kits/100-day-kit
- Autism Navigator
  - Family Resources and glossary of videos of early symptoms in toddlers
  - http://www.autismnavigator.com
- CDC “Learn the Signs. Act Early” Web site provides free resources to help families recognize developmental concerns, including autism
  - https://www.cdc.gov/ncbddd/actearly



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- Request for Information: Input on Future Directions for the Science of Behavior Change Common Fund Program (NIH; 02/20/14)
- Massage Therapy for Health Purposes: What You Need To Know
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- Chronic Pain and Complementary Health Approaches: What You Need To Know
- Credentialing: Understanding the Education, Training, Regulation, and Licensing of Complementary Health Practitioners
- Blog Post: Fast-Forwarding Natural Products Research (01/24/14)