

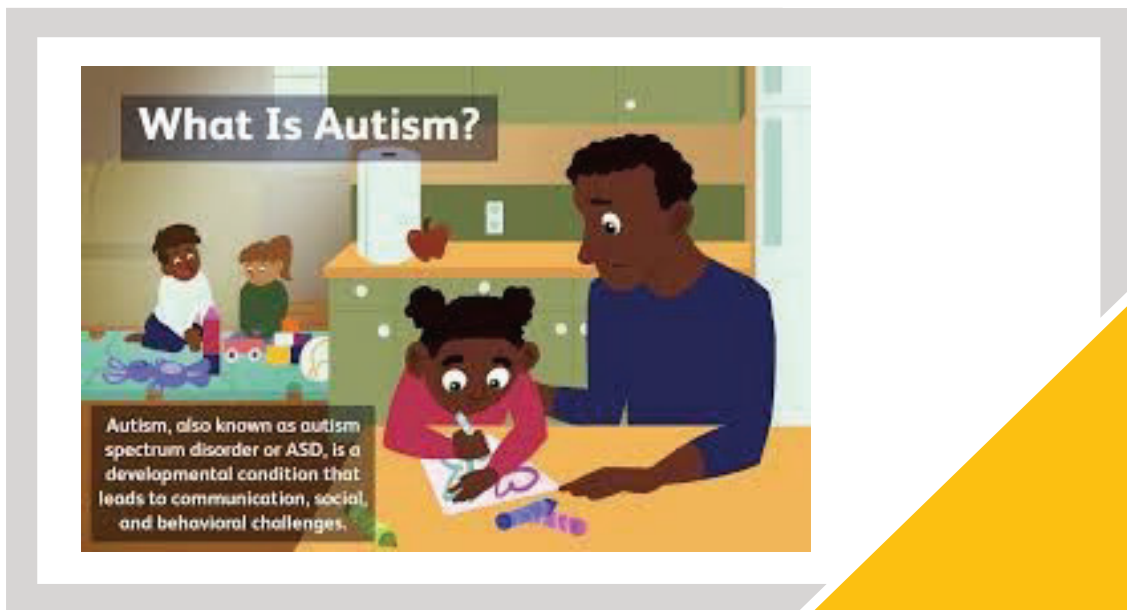


Autism from a
Pediatric
Primary Care
Provider's
Perspective

Journey into my world: Let the Adventure Begin!

Nina Ford Johnson, MD
April 27, 2024





DSM V definition of Autism

- A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history (examples are illustrative, not exhaustive):
- A. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and **failure of normal back-and-forth conversation**; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
 - B. Deficits in nonverbal communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to **abnormalities in eye contact** and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
 - C. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to **absence of interest in peers**.

Why is knowing these facts important?

8 years old: 1 in 36 children have a diagnosis of autism spectrum disorder as of 2020 (in 2014: 1 in 59)

More than 5 million people in the US

The ethnicity factor has changed over the decades (minority diagnoses have increased over Caucasian rates)

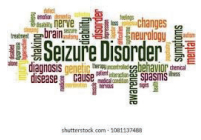
In my practice: ~59 children with autism (80% of those are males)

DSM V definition of Autism

- B. Restricted, repetitive patterns of behavior, interests, or activities, as manifested by at least two of the following, currently or by history (examples are illustrative, not exhaustive):
 - B. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., simple motor stereotypes, lining up toys or flipping objects, **echolalia**, idiosyncratic phrases).
 - C. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or **eat same food every day**).
 - D. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g., strong attachment to or **preoccupation with unusual objects**, excessively circumscribed or perseverative interests).
 - E. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment (e.g. apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, **visual fascination with lights** or movement).

Gold Standard for Development

- ASQ (ages & stages questionnaire)
 - 12-15 months (any abnormalities): refer to Early Intervention
 - 18-24 months: ASQ and MCHAT (modified checklist for autism in toddlers): EIS and Developmental referral
 - Plus ST, OT, PT referrals (ABA after formal autism diagnosis)
 - 3yr-4yr: it's time to go to school!!! (or full-time ABA, but pickings are slim)
 - 4yr+: school





ASQ 12 month

Davis, Chazalah David Scan on 1/27/2020 by User: E461082 of 12 Month ASQ

ASQ-3 12 Month Questionnaire page 1 of 6

OVERALL
Parents and providers may use the space below for additional comments.

- Does your baby use both hands and both legs equally well? if no, explain: YES NO
- Does your baby play with sounds or seem to make words? if no, explain: YES NO
- When your baby is standing, are her feet flat on the surface most of the time? if no, explain: YES NO
- Do you have concerns that your baby is too quiet or does not make sounds like other babies do? if yes, explain: YES NO
Absolutely Not!
- Does either parent have a family history of childhood deafness or hearing impairment? if yes, explain: YES NO

E101120500 Age 8 Steps Questionnaire, Third Edition (ASQ-3), Brookes & Baker © 2007 Paul H. Brookes Publishing Co. All rights reserved.

Printed by Nina Ford Johnson, MD on 4/20/24 8:06 AM Page 5 of 6

ASQ 18 month

4/20/24, 8:07 AM Davis, Chazalah David (MR # 6000038300)

ASQ-3 18 Month Questionnaire page 3 of 6

OVERALL
Parents and providers may use the space below for additional comments.

- Do you think your child hears well? if no, explain: YES NO
Not completely sure
- Do you think your child talks like other toddlers his age? if no, explain: YES NO
Very few words. Has stopped using some that he was using before (da-da and bye-bye)
- Can you understand most of what your child says? if no, explain: YES NO
He says ba-ba for ball and ma-ma
- Do you think your child walks, runs, and climbs like other toddlers her age? if no, explain: YES NO
He just started walking in the last 2 weeks
- Does either parent have a family history of childhood deafness or hearing impairment? if yes, explain: YES NO
- Do you have concerns about your child's vision? if yes, explain: YES NO

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Davis, Chazalah David (MR # 6000038300) Printed by Nina Ford Johnson, MD (DR01417) at 4/20/2024 8:07 AM 5/7

MCHAT
www.m-chat.org

Child's name Chazalah Davis Date 2/9/21
Age 2 Relationship to child mother

MCHAT-R™ (Modified Checklist for Autism in Toddlers - Revised)

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	<input type="radio"/> No
2. Have you ever wondered if your child might be deaf?	<input checked="" type="radio"/> Yes	<input type="radio"/> 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	<input checked="" type="radio"/> No
4. Does your child like climbing on things? (FOR EXAMPLE, furniture, playground equipment, or stairs)	<input checked="" type="radio"/> Yes	<input type="radio"/> 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	<input checked="" type="radio"/> 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)	<input checked="" type="radio"/> Yes	<input type="radio"/> 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	<input checked="" type="radio"/> No
8. Is your child interested in other children? (FOR EXAMPLE, does your child watch other children, smile at them, or go to them?)	<input checked="" type="radio"/> Yes	<input type="radio"/> 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)	<input checked="" type="radio"/> Yes	<input type="radio"/> 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?)	<input checked="" type="radio"/> Yes	<input type="radio"/> No
11. When you smile at your child, does he or she smile back at you?	<input checked="" type="radio"/> Yes	<input type="radio"/> No
12. Does your child get upset by everyday noises? (FOR EXAMPLE, does your child scream or cry to noises such as a vacuum cleaner or loud music?)	<input checked="" type="radio"/> Yes	<input type="radio"/> No
13. Does your child walk?	<input checked="" type="radio"/> Yes	<input type="radio"/> 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?	<input checked="" type="radio"/> Yes	<input type="radio"/> No
15. Does your child try to copy what you do? (FOR EXAMPLE, wave bye-bye, clap, or <u>blow</u> make a funny noise when you do)	<input checked="" type="radio"/> Yes	<input type="radio"/> No
16. If you turn your head to look at something, does your child look around to see what you are looking at?	Yes	<input checked="" type="radio"/> 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	<input checked="" type="radio"/> 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	<input checked="" type="radio"/> 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	<input type="radio"/> No
20. Does your child like movement activities? (FOR EXAMPLE, being swung or bounced on your knee)	<input checked="" type="radio"/> Yes	<input type="radio"/> No

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MCHAT

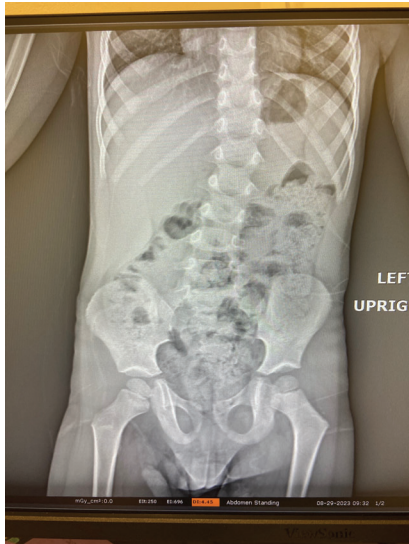
2 yr

Evan



Evan: GI

- Pica (2 years of age) – blanket, cardboard, paint, fragments of bricks
 - Lead screen was in normal range; Hb is 13 with normal MCV
- He has a "high pain tolerance" per mom
- Diarrhea with gluten foods at age of 3 years
- Diet: pizza, bread/toast, chicken and rice, chips, dried fruit
- 11/2022: first admittance to the hospital for bowel flush secondary to constipation (was very pale in my office, vomiting bile). He required a NG tube for evacuation
- Several ER trips: August-September 2023; last hospitalization was in Sept 2023 for another bowel cleanout



Feeding disorders

Food selectivity: based on texture, color, temperature

Behavioral refusal: holding food in the mouth, gagging, and emesis

Pica

Rumination

Other considerations: GE reflux, **dental pain**, food allergies, lactose intolerance, constipation

Abrian

- Increased risk for seizures in those with ASD
- Rate of seizures among people with ASD has been reported to range from 7-23%
- Genetic disorders associated with ASD may contribute to early seizure risk in early childhood
- 9 month old checkup: episodes of dropping his head, arms are straight out, and this will lift his head and then laugh (occurring for over a month); can happen back-to-back
- 12 months: abnormal EEG (diffuse background slowing, more over the temporal regions) and referred him for autism evaluation

Abrian

- Treated for infantile spasms and started on steroids (12 months)
- My 12 month old exam: smiles, moans frequently; not crawling, but trying to stand, plays with his fingers in front of his face
- Treatment: weaned off of steroids and put on Topamax
- Genetics: KMT2E-associated neurodevelopmental disorder aka O'Donnell-Luria-Rodan syndrome
 - Fewer than 100 cases reported in the literature (fairly new syndrome)
 - Some characteristics: 69% with motor delay, 83% speech delay, 31% autism, 15% epilepsy, intellectual disability 72%

Fragile X

- X-linked disorder; loss-of-function in the FMR1 gene due to the unstable expansion of CGG repeat at that specific gene
- Most frequent form of inherited ID
- Males > females
- Phenotype: long and narrow face with prominent forehead and chin, large ears
- 18-67% with FXS meet the criteria of ASD
 - Males with FXS & ASD: have issues of social interaction, academic achievement, and are more than likely to have seizures

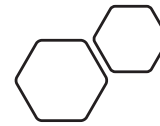
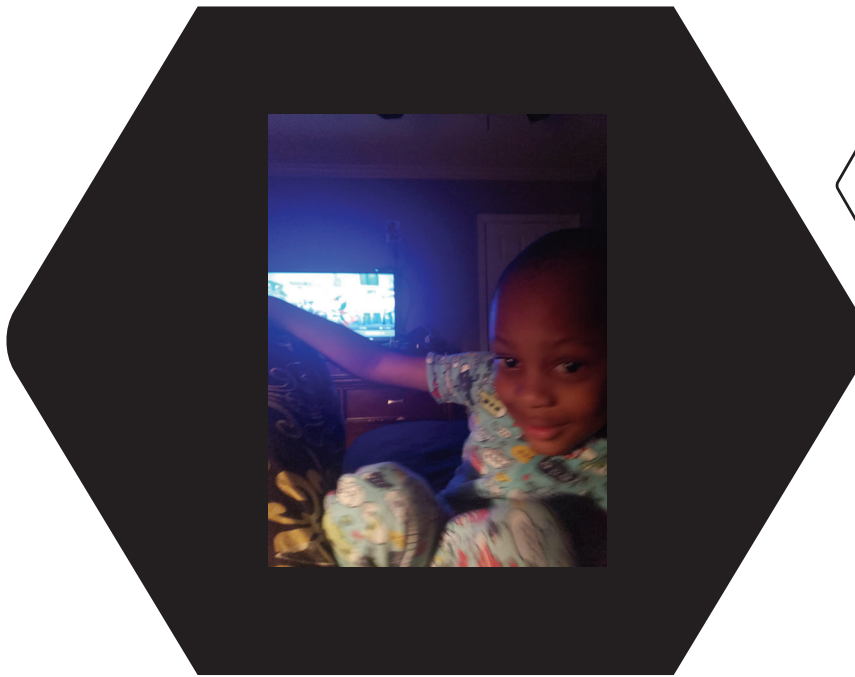




Chaz

Chaz

- Has never wanted to go to sleep at night
- Routine: wake up at 3:30am and will not return back to sleep until 1am the next day (maybe a nap at school); happens several nights a week
- Melatonin helps him go to sleep, but not stay asleep
- I tried Clonidine back in Feb 2023, but mom said it made it “worse”



insomnia

- Problems with initiating and maintaining sleep are reported for 50%—80% of children with ASD
- Why? Differences of melatonin metabolism, disruption of neurotransmitter systems, restless leg syndrome, anxiety, epilepsy, ADHD, or motor disorders
 - Medical disorders: sleep apnea, asthma, eczema
- Treatment: melatonin (1-6mg), clonidine, **antihistamines**
- **Hydroxyzine**

Maliah

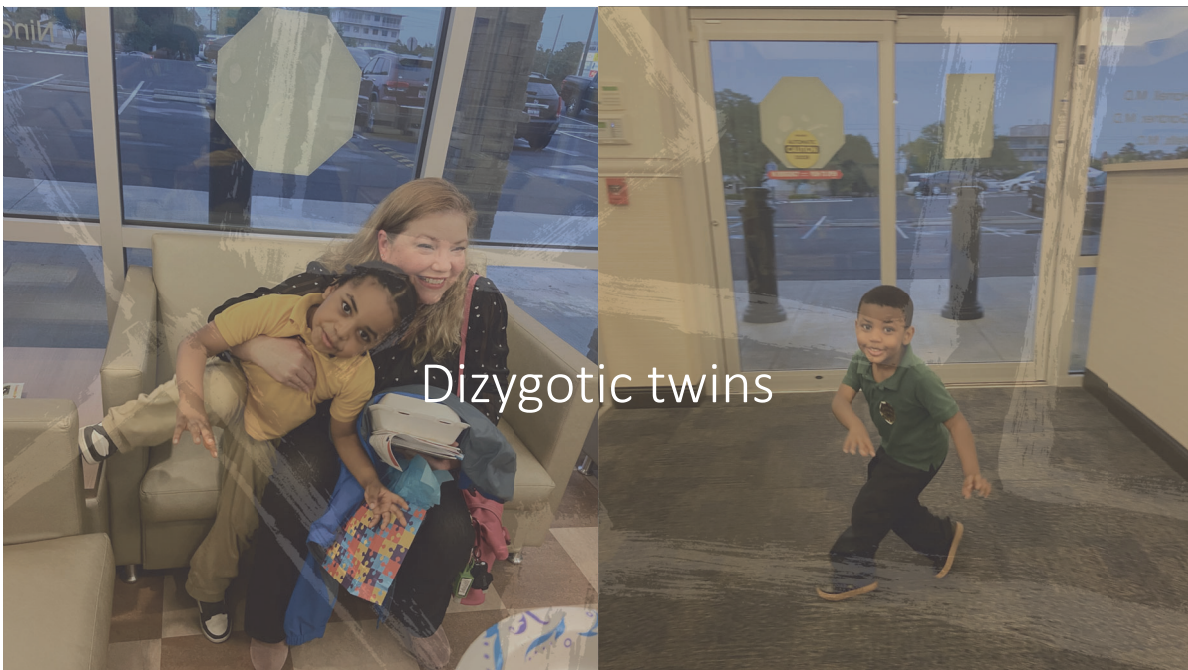


Aggression

- Co-occurring behavioral symptoms: hyperactivity +/- inattention, aggression, outbursts, and self-injurious behavior
- 70-90% of children with ASD have psychiatric conditions (ADHD, anxiety, OCD, mood disorders, conduct disorders etc..)
- Maliah (at the age of 4 years)
 - Hitting (“beating up”) her older sister
 - Throwing her shoe at the teacher; biting and hitting her teacher/classmates
 - Running in front of cars
 - Treatment: Risperidone
 - Now: Maliah is 6 years old and won student of the month at her school for March

ADHD

- More than $\frac{1}{2}$ of children with ASD also have ADHD (hyperactive)
- *up to $\frac{1}{4}$ of children with ADHD have low-level signs of ASD (difficulty with social skills, problems with texture)*
- Some of my favorite medications: Quillivant, Focalin XR, Vyvanse
- Side effects: appetite, emotional lability, unmasking of other comorbidities (anxiety, depression etc...)



Dizygotic twins

An analysis involving over 6400 twin pairs revealed a 53-67% concordance in dizygotic twins (98% in monozygotic twins)

Siblings, of those twins, may be at risk for symptoms related to ASD that do not meet the threshold for a diagnosis of ASD, but similar phenotype

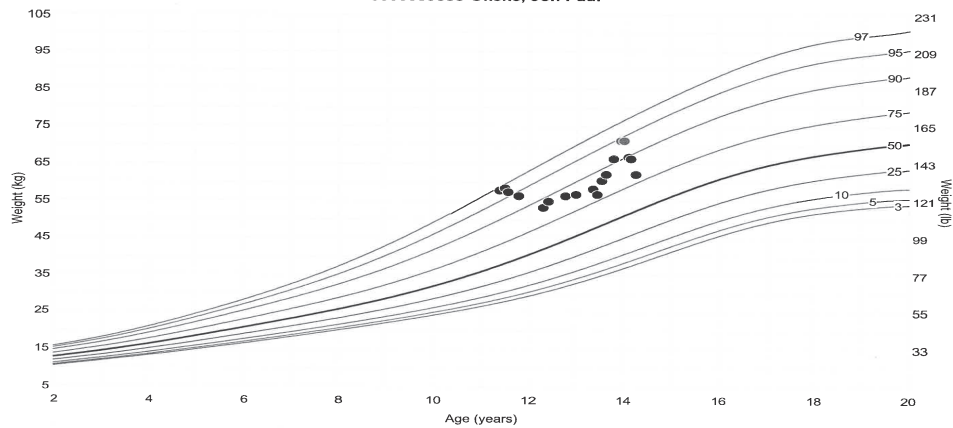
My practice: 7 twin individuals (one child not diagnosed with autism, but who still has autistic features to me)



Jon Paul



Growth Chart: United States
Weight-for-age Percentiles (Boys, 2 to 20 years)
6000039388-Okeke, Jon Paul



Jon Paul

- 12/2023: intractable headaches (had to be admitted to USA)
 - During this hospital visit, he disclosed to intern that why he was having headaches is because he was not eating
- Started several years ago after watching videos of the fat acceptance movement; which made him fearful of gaining weight
- 11/2023-2/2024 = 16lb weight loss (intentional, restricting)
- Accompanying comorbidity: depression (therapy, Prozac)
- Sexuality



Causes/observations

- Environmental factors (like asthma): examine ASD and population-level associations for organophosphates, pesticides, metals, volatile organic compounds, and air pollution (particulate matter and nitrogen dioxide)
- Association between trauma (parental) and ASD
- Observation: very few of my kids who were put on specialized formulas, have ASD



Setbacks or why does it take long to diagnose kids?

- Parents of children with ASD report more stress (more than ½ my children are lead by single-parent households..with more than one child who has a ASD)
- Increased costs
- More time off of work
- Older children come to me from other PCPs where screening wasn't available
- Vaccines are not the problem
- Language barriers



Thank you!!!!

Dr. Mark Haygood

Zack Johnson
(tech support)

My beautiful
patients and their
sweet families

You!!!!