2nd Annual NorthShore Pediatric Symposium

BETTER UNDERSTANDING THE SPECTRUM OF AUTISM: WHY AND WHEN IT MATTERS

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Disclosure Information Susan Fielkow, M.D. 2nd Annual NorthShore Pediatric Symposium

- I have no financial relationships to disclose
- I will not discuss off label use and/or investigational use in my presentation

Autism Spectrum Disorder 299.00 (F84.0) DSM-5

- Diagnostic Criteria
- 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, see text):
- 1. **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.

Autism Spectrum Disorder- DSM-5

- 2. **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.
- 3. **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.

Specify current severity:

 Severity is based on social communication impairments and restricted repetitive patterns of behavior (see Table 2).

- 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; see text):
- 1. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases).
- 2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns or verbal nonverbal behavior (e.g., extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat food every day).

- 3. **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 interest).
- 4. Hyper- or hyporeactivity to sensory input or unusual interests 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).

Specify current severity:

 Severity is based on social communication impairments and restricted, repetitive patterns of behavior (see Table 2).

- C. Symptoms must be present in the early developmental period (but 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. These disturbances are not better explained by intellectual disability (intellectual developmental disorder) 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.

(protects earlier diagnoses)

- Note: Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger's disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder. Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.
- Table 2 Severity levels for autism spectrum disorder

ASSESSMENT TOOLS

- MCHAT-R
- DSM-5 criteria
- SRS Questionnaire
- CARS-2
- SRS
- ADI-R
- ADOS

M-CHAT-R/F (Modified Checklist for Autism in Toddlers, Revised with Follow-Up)

- 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?)
- 2. Have you ever wondered if your child might be deaf?
- 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?)
- 4. Does your child like climbing on things? (FOR EXAMPLE, furniture, playground equipment, or stairs)
- 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?)
- 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)
- 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. This is different from your child pointing to ASK for something [Question #6.])
- 8. Is your child interested in other children? (FOR EXAMPLE, does your child watch other children, smile at them, or go to them?)
- 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)

- 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?)
- 11. When you smile at your child, does he or she smile back at you?
- 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?)
- 13. Does your child walk?
- 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?
- 15. Does your child try to copy what you do? (FOR EXAMPLE, wave bye-bye, clap, or make a funny noise when you do)
- 16. If you turn your head to look at something, does your child look around to see what you are looking at?
- 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"?)
- 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"?)
- 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?)
- 20. Does your child like movement activities? (FOR EXAMPLE, being swung or bounced on your knee)

MCHAT- R

- Great screening tool
- Need to consider DEVELOPMENTAL AGE
- If screening is "positive," should discuss reported items of concern
- Refer for further evaluation don't delay due to age

WHEN TO DIAGNOSE/WHEN TO REFER?

- Depending on comfort level of PCP and/or availability of resources
- Need confirmation or unsure of diagnosis
- Clarification between school and medical diagnosis
- Need assistance with management:
 - Coordination of care: schools, therapists, medical providers
 - Community resources
 - Medical evaluation

THE &UTISM SPECTRUM IS BRO&D &ND COMPLEX

CONCERNS REGARDING OVER DIAGNOSIS CONCERNS REGARDING UNDERDIAGNOSIS

SCHOOL DIAGNOSIS MEDICAL DIAGNOSIS WHY DO THEY DIFFER?

School Classification of Autism

- School psychologists and medical professionals don't always interpret autism spectrum criteria the same way
- School classification may provide helpful interventions to improve school participation
- Medical diagnosis needed to obtain specific private therapies
- PCP has an important role in clarifying differences in diagnostic processes and why/why not the child meets criteria for an autism spectrum disorder
- May need further evaluation to clarify

WHY DOES IT MATTER?

- Taxonomy
- Consistency and consensus
- When we call it a duck...
- Access to appropriate interventions
- Parents, teacher, providers, therapists better understand the developmental-behavioral profile

Is he too young to diagnose?

- Toddlers can be diagnosed
- Must demonstrate social and communication deficits that are significantly lower than cognitive skills

Early Communication and Social Skills

- Social smile
- Caregiver preference
- Upset by caregiver leaving (object permanence)
- Vocal tennis
- Imitative babbling

Joint Attention

Gaze monitoring
 8-9 months

Follow a point 10 months

Point to request
 12 months

protoimperative: Baby wants you to see what she/he sees

Point to comment 14-15 months

protodeclarative: Baby wants you to see what he/she sees

ADOS-2 Toddler Version

- Under 30 months of age
- Cognitively at least 12 months

When it looks like a duck but isn't

 Child who presents with excitable motor movements (arm waving, tapping), restricted interests (Mine Craft, Pokemon), and struggles with social interaction, and doesn't answer questions in class, except with simple responses

High functioning Autism Spectrum

Can look like:

- Disrespectfulness
- Offensive to others
- Inattentive behaviors
- Social anxiety
- ADHD
- Just too smart to fit in

And vice versa

How can you tell

Try to determine the difference between

Not getting it and Not doing it

CASE PRESENTATIONS

J.G.

J.G. is a 22 mo boy born to a 34 year old father and 36 year old G2P1 mother

Delivered at 37 weeks, via uncomplicated repeat cesarean section

Birth weight: 6 lb 6 oz

APGARS: 8 at 1 min., 9 at 5 min.

Mild jaundice

Newborn hearing screen normal

Physical Exam

Normal growth parameters and head circumference

Hearing done by Early Intervention: normal

Vision: not tested

No cutaneous findings

Family history: two cousins with autism

Development

- FM: can't use spoon or fork
- No scribble
- Holds bottle, but will not use cup
- Drops several blocks in a cup.
- Pushes buttons with index finger, twists door knobs, opens lids

Development continued:

- GM: Sat 8-9 mo
- Crawled around 1 year
- Walked: 15 months
- No running, jumping.
- Crawls up stairs

Speech/Communication

- Babble with vowel-consonant combinations in response to videos
- Says "mama, dad, yes, no, cat, bye" but not to name or call someone. Will say "bye" in appropriate context at times
- Will not repeat words on request
- No protoimperiative or protodeclarative point
- No nod, head shake, wave. No gesture to make choice
- Brings toy or object to get help

Social

- Makes eye contact
- Show interest in others but doesn't interact
- Rarely responds to his name
- No showing or initiation of joint attention
- Occasional response to joint attention
- Very affectionate with family members, seeks comfort, likes to cuddle
- Cries with strangers

Play

- Cars: rolls, pushes and chases them
- Throws balls back and forth with a person
- Plays some games on the tablet- drags food and feeds people. Matches shapes. Finds Utube videos.
- No pretend with toys or objects

Restricted/Repetitive Behaviors or Interests

- Hand flaps, claps repetitively
- High pitched scream with fist clenching/pulls at hands in response to frustration or aversive stimuli

Sensory

- Smells mom's neck
- Will not eat many solid foods, except chips
- Doesn't like to chew things
- Not visual, tactile, auditory sensitivities

Problem or not?

- MCAT: several items of concern
- History: concerns regarding interaction but reassuring items: likes to cuddle, makes eye contact, better interaction with some familiar individuals- throws ball back and forth, brings objects to get help, rolls cars back and forth.
- Important: Plays some games on the tabletdrags food and feeds people. Matches shapes. Finds U-tube videos.

ADOS- 2 TODDLER VERSION

 Language and Communication: JG used one word "Hi" during the assessment. He often used open vowel sounds while playing ('Ahhhh" and "oooo"). He directed some of his vocalizations toward the examiner or toward his father to indicate interest in an activity. At times his cries were odd, and seemed to occur out of context. His language was too limited to note echolalia or any stereotyped speech. He did not use another person's body as a tool. He did not point or use any gestures during this assessment.

Reciprocal Social Interaction:

J.G did not use eye contact to modulate social interaction. He did not make eye contact during the teasing toy play. He had few facial expressions and did not direct expressions toward the examiner or toward his father. He showed occasional pleasure in interaction, notably during the ball play. He did not respond to his name with eye contact. He did not make bids for attention when ignored, he was content playing independently. He did not request, even when prompted. He seemed to enjoy the bubbles, but when asked if he wanted more, he looked at the bubbles, but made no attempt to request or communicate with me. He did not give objects. He did look at his father on several occasions when he seemed to be enjoying an activity. He did not make clear social overtures to me or to his father. He was not consistently engaged, and rapport was not sustained.

- Play: JG played with the ball, the car, the pop-up toy, the music box and the bubbles. He did not engage in representational play other than pushing the car. He did not imitate or play with imagination.
- Restricted, Repetitive Behaviors or Interests: JG bent down to watch the wheels turn when pushing the car. He covered his ears several times without an obvious stimuli. He engaged in excitable motor mannerisms (flapping). He engaged with the balls and the pop-up toy to an unusual degree, but was able to move on when those items were removed.

ADOS-2 RESULTS

- Social Affect Total: 16
- Restricted and Repetitive Behavior Total: 5
- Total: 21
- ADOS-2 Range of Concern: moderate-tosevere concern

L.M.

- L.M. is an 11 –year old boy with a history of speech/language delay, specific learning disability (language), anxiety, and social problems. Spanish is primary language at home
- Ongoing school concerns regarding lack of academic progress and possible autism. Rating scales done by school teacher, SLP, SW endorsed "very likely" symptoms of autism on the GARS (Guilliam Autism Rating Scale) and on the BASC: Atypicality, Anxiety, Withdrawn and some ADHD

L.M.

- L.M. rarely talks to or interacts with peers, struggles to answer questions, often answers with the same inappropriate phrases, poor conversation, and talks to himself.
- Receptive language: 1st percentile
- Expressive language: 2nd percentile

L.M.

Parents do not see same behaviors and note that L.M. is very funny, interactive, attention seeking at home. He has trouble expressing himself. He is very shy and won't to talk to people with whom he is unfamiliar

L.M. - ADOS-2

- Social Affect Score: 3
- Restricted and Repetitive Behavior: 0
- (Autism spectrum cut-off=7, Autism cut-off=9)
- Noted limited eye contact, grammatical language errors and some unusual word choices.

NEPSY-II

- Affect Recognition subtest: Scaled score = 8
- Theory of Mind: 2nd percentile

L.M. follow up

- Treated with Prozac and continued social work support at school
- Happy, engaged in class
- Teacher complimentary of his participation
- Actively playing outside with neighborhood kids

High functioning Autism Spectrum

Can look like:

- Disrespectfulness
- Offensive to others
- Inattentive behaviors
- Social anxiety
- ADHD
- Just too smart to fit in

And vice versa

Tools

Asperger Syndrome Diagnostic Scale DSM-5 Criteria

- W.C. initially presented with a neuropsychological diagnosis of ADHD-CT and ODD
- He demonstrated difficulty with flexibility and low frustration tolerance.
- Child is very bright and frequently appeared to be sarcastic and manipulative, however his mother patently disagreed with this impression

Asperger's Syndrome Diagnostic Scale

LANGUAGE/COMMUNICATION

- 1. Speaks like an adult in an academic or "bookish" manner, or sounds like a "little professor." May overly use correct grammar or mature vocabulary. :sometimes
- Talks excessively about a favorite, or unusual topics that hold limited interest for others: Yes, his interests change but becomes very interested in things and talks frequently about current interest
- 3. Uses words or phrases repetitively: yes, "booya", "moola"
- 4. Does not understand subtle jokes, e.g., sarcasm: **He can be sarcastic** and loves jokes
- 5. Interprets conversations or statements literally. Doesn't understand metaphors, idioms, figures of speech: He is good at metaphors

- 6. Has peculiar voice characteristics (i.e., sing-song, monotone, accents, inappropriate volume or rate): sometimes feedback from school is that his speech is atypical in volume and rate
- Acts as though he/she understands more that he/she does, without really understanding maybe
- Frequently asks inappropriate questions (i.e. out of context, or socially inappropriate): yes example "what is 7+7" when asked if he has questions here at appointment
- 9. Difficulty beginning and continuing conversations (i.e. trouble with back and forth exchanges) : **Yes**

SOCIAL BEHAVIORS

- 1. Uses few gestures while speaking. Lacks body language: **No**
- Avoids or limits eye contact : sometimes, (exacerbated by anxiety)
- 3. Has trouble relating to others, not easily explained by shyness, attention, or other things : **Yes**
- 4. Demonstrates few or inappropriate facial expressions (i.e. flat or exaggerated affect): **Yes exaggerated**
- 5. Shows limited or no interest in peers : **limited he does have** some interest in friends, but he is selective
- 6. Prefer to be with adults more that peer: **no**

- 7. Has few or no friends, despite a desire to have them: **He has a few friends**
- 8. Has difficulty making or keeping friends : **He can make friends**, **but it is hard to keep them**
- 9. Does not respect others' personal space. : **yes**
- 10. Shows little interest in what others say or find interesting : no
- 11. Has trouble understanding the feelings of others; why others would feel a certain way: **no**
- 12. Does not understand or use rules governing social behavior: **Yes, this causes difficulty**
- 13. Trouble understanding social cue: **Yes, this causes difficulty**

MALADAPTIVE BEHAVIORS

- Does not change behavior to match the environment (e.g. notices others in the room are quiet and adjusts his voice accordingly): Yes
- 2. Engages in inappropriate behavior related to obsessive/favorite interest : **No**
- 3. Antisocial behavior : sometimes
- 4. Exhibits strong reaction to change in routine : No
- 5. Becomes anxious or panics if unscheduled/unanticipated event occurs : **sometimes**

- 6. Appears depressed : **yes, sometimes**
- 7. Demonstrates repeated, obsessive or ritualistic behavior : **No**
- 8. Displays immature behaviors : Yes
- 9. Frequently loses temper or has tantrums : **Yes**
- 10. Frequently seems overwhelmed, especially in crowds or demanding situations : **Yes**
- 11. Imposes narrow interests, routine or structures on others : **No**

COGNITIVE FEATURES

- 1. Displays a superior ability in a restricted area, but has average to above average skills in other areas : **No, he is above average in several areas**
- 2. Has extreme or obsessive interest in narrow area or subject : He will get into one topic intensely for a short period of time, and then move on to something else
- 3. Functions best when engage in familiar and repeated tasks: No
- 4. Has excellent memory : **No, he has problems with working memory**
- 5. Learns best through pictures or written words, as opposed to verbally: **Yes, written words**

- Average to above average intelligence (not including specific learning disabilities): yes
- 7. Aware that he/she may be different from others: Yes
- 8. Oversensitive to criticism. May misinterpret minor corrections or instructions as criticism. : **Yes, he is very sensitive**
- 9. Lacks organizational skills; Yes
- 10. Lacks common sense; Yes

SENSORY/MOTOR

- 1. Unusual or over-reaction to loud, unpredictable noises, or even some everyday noises (e.g., screams, covers ears, withdraws); **Yes, he is sensitive to noise, particularly when he is concentrating**
- 2. Stiffens, flinches or pulls away when hugged : depends on who hugs him
- 3. Overreacts to smells that are hardly noticed by others: Yes, he is sensitive to smell
- 4. Prefers to wear clothing made of certain fabrics, or is overly sensitive to the feeling of things: **He used to be, but he is more flexible now**
- 5. Restricted diet consisting of same foods: **Yes**
- 6. Trouble with handwriting or other fine motor tasks (i.e., buttoning, typing, snapping): **Yes**
- 7. Clumsy or uncoordinated; **somewhat**

W.C.- ADOS-2

W. C. used sentences in a largely correct fashion, including complex speech. He occasionally used an exaggerated intonation, particularly when reading the book and interpreting the cartoon. He did not echo speech. His use of words was more formal than most individuals at the same level of expressive language (he described happiness as "pleasure and satisfaction coursing through my veins"). He occasionally offered information about his own thoughts feelings or experiences ("I do not like action figures." "I prefer legos"). He asked about my thoughts, thoughts, feelings and experiences only when given specific prompts. He was able to report events with reasonable detail when asked a general question. His speech included some spontaneous elaboration of responses, but conversation was limited in flexibility, and when he seemed stumped, he would ask about drinks ("do you like Coke or Pepsi?").

 W.C. made eye contact, and directed facial expressions, but they were often exaggerated. His play was primarily self-directed, and while he seemed somewhat eager to please, did not seem to take genuine pleasure in social interaction. W.C. did spontaneously communicate understanding of emotions in several characters in pictured stories. He showed limited insight into social situations and relationships. When describing his friends he was able to name characteristics about them that he liked, "He loves puns, She is very smart, he likes to draw"), but also mentioned that "She is not as smart as I am" and, "He does not draw as well as me." He struggled when describing the positive aspects of marriage ("my DNA gets carried on").

His response to a coping with a bully was limited and immature. He commented that he would hit the bully, or make up a snappy response such as telling him he has toilet paper on his shoe. His social overtures were slightly awkward, and he inquired several times about my cola preference. He did make frequent attempts to get my attention. He responded to social overtures, but responses were often awkward, exaggerated or limited to his interests. Overall, rapport was comfortable at times but not sustained, as W.C.'s behavior seemed inappropriate at times. (At times his affect was exaggerated, at times he was overly formal, and when he called the frog a turtle in the book, and I gently noted his error he hit himself on the head several times and shouted "damn! damn!")

- Play: W.C.'s play was limited in range. He was able to use objects for something other than their intended purpose, but could not use action figures. He was unable to create a story. He used the objects to create a short space fight, and then stopped, and said that it was over.
- He made occasional references to beverages (Coke, Pepsi, highly caffeinated coffee and smoothies). This occurred to an unusual degree and these comments occurred out of context

ADOS results

ADOS-2 total score: Autism Spectrum Disorder

- Social Affect Total: 6
- Restricted and Repetitive Behavior Total: 2
- Total: 8 (Autism cut-off = 9; Autism spectrum cutoff = 7)
- ADOS Classification: Autism Spectrum
- ADOS-2 Comparison Score: 5, corresponding to a moderate level of autism spectrum-related symptoms

BODY LANGUAGE-WITHOUT WORDS

BY OTTERDISASTER











