



# Autism Spectrum Disorders for the Primary Care Practitioner and Other Providers

Friday, May 5, 2023

*Virtual Conference*

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Downstate Health Sciences University



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## Early Childhood Development/ Autism Spectrum Disorders

May 5, 2023

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I have nothing to declare (except I could use more time)



What is Infant & Early Childhood Mental Health?

**The developing capacity from birth to 6 “to experience, regulate, and express emotions; to form close relationships; and to explore the environment and learn”<sup>1</sup> — all in the context of family, community, and cultural expectations for young children.**

An infant, toddler and young child’s mental health is every part as important as their physical health. Mental health matters for the growth and maturity of the brain and body and for the social and emotional development of a person — now and for the whole lifetime.

<sup>1</sup> The Center on the Social Emotional Foundations for Early Learning. Infant Mental Health and Early Care and Education Providers. Vanderbilt University, retrieve from: [http://csefel.vanderbilt.edu/documents/rs\\_infant\\_mental\\_health.pdf](http://csefel.vanderbilt.edu/documents/rs_infant_mental_health.pdf)

#### What is Early Relational Health?

Early Relational Health is the state of emotional well-being that grows from the positive emotional connection between babies and toddlers and their parents/caregivers when they experience strong, positive, and nurturing relationships with each other. Early Relational Health is foundational to children’s healthy growth and development and their parents’/caregivers’ sense of competence, connection, and overall well-being. These resilient and enduring relationships also help to protect the family from the harmful effects of stress.



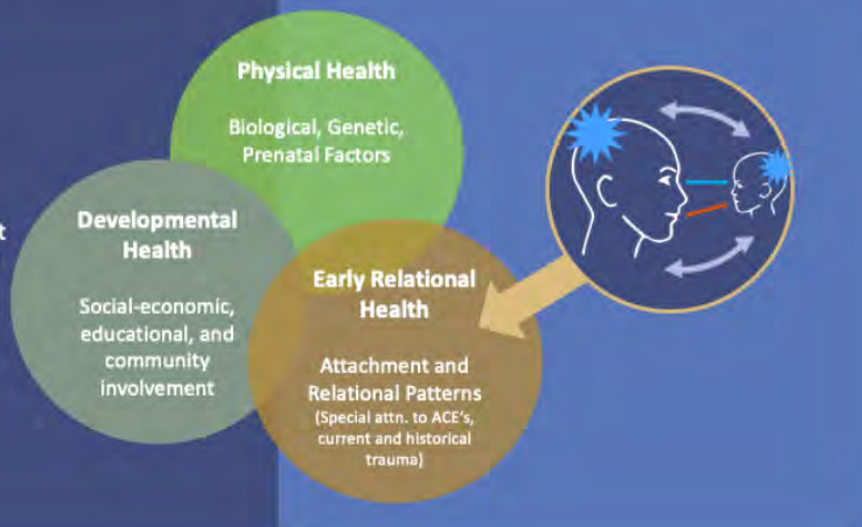
**Suggested citation:** Center for the Study of Social Policy (2022). *How to Communicate Effectively About Early Relational Health: What It Is and Why It Matters A Messaging Guide*. Retrieved from <https://cssp.org/>



## Early Relational Health

### Early Relational Health

is a multi-dimensional and dyadic construct established by the caregiver-child interactions during the First 1000 Days of life that build lifelong health, early learning, social-emotional capacities, self-regulation and resiliency.



[www.CSSP.org](http://www.CSSP.org)

David W. Willis, MD FAAP

Senior Fellow  
June 12, 2019

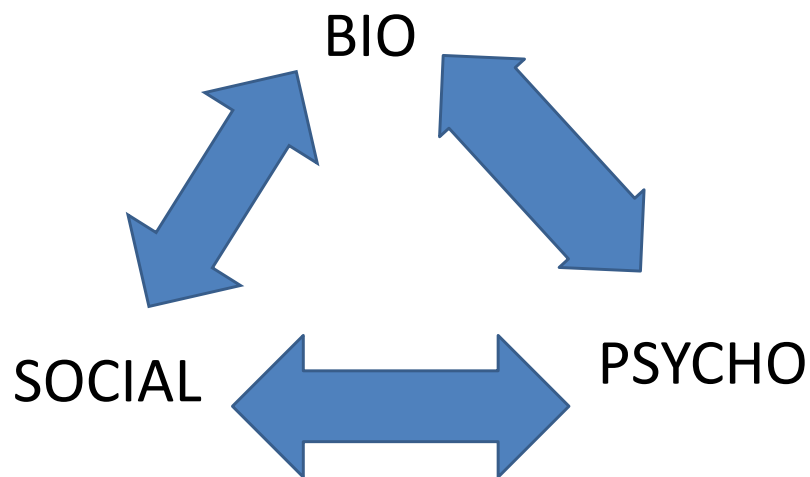
Culturally “normal”  
and/or healthy and/or  
positive growth  
and/or development  
is not a given

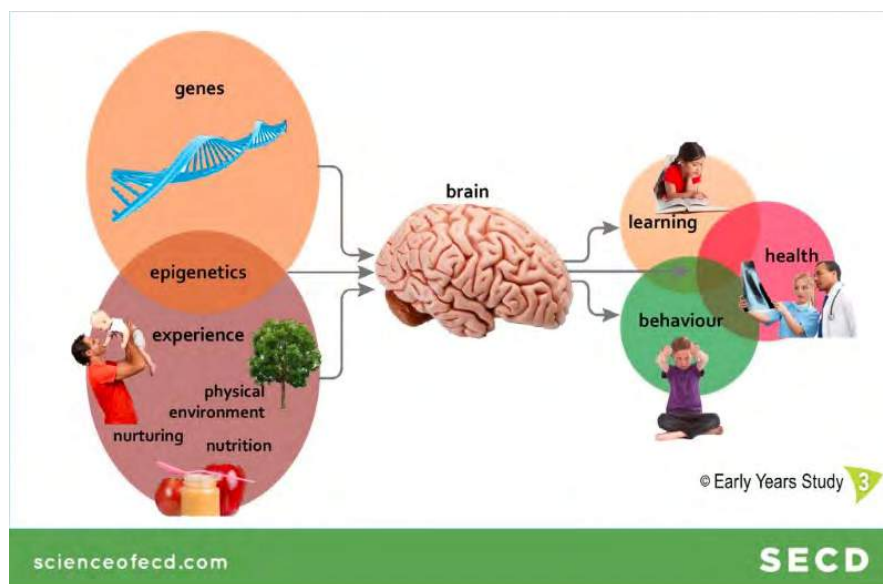
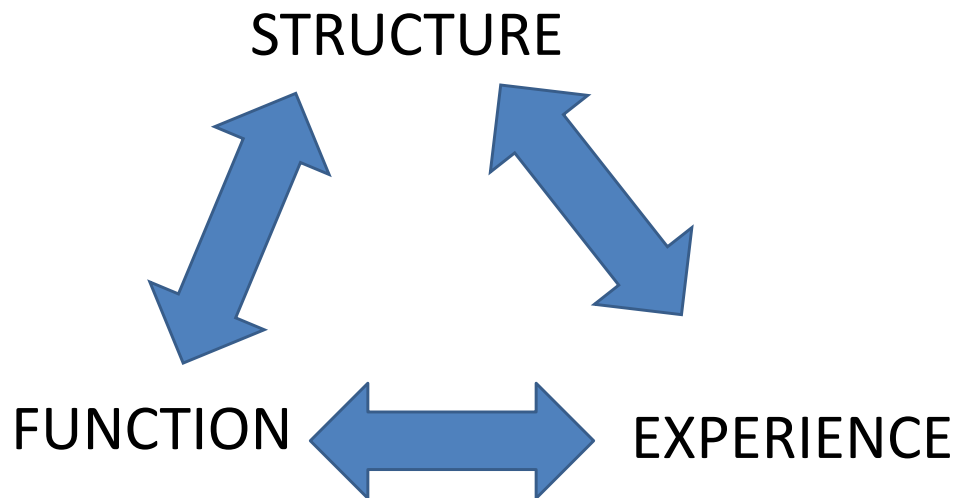


Nor is it static

Development is always an ongoing dynamic  
and  
the ongoing dynamic is always developing



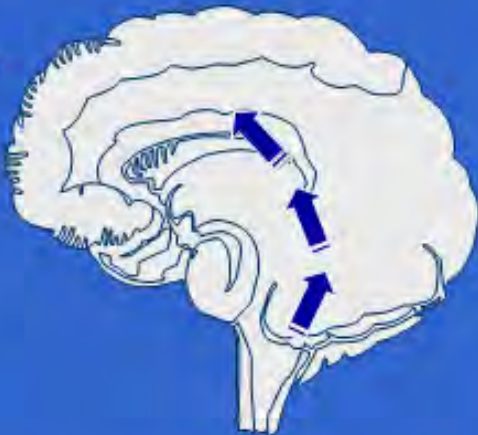






## Experience Grows the Brain

- Brain development happens from the bottom up:
  - From primitive (basic survival: brainstem)
  - To more complex (rational thought, planning, abstract thinking: prefrontal cortex)



Source: Grilo, C. K., Lutz, D. K., & Foster Care Subcommittee of the Child Welfare Committee, National Child Traumatic Stress Network, (2010). Caring for children who have experienced trauma: A workbook for resource parents. Los Angeles, CA and Durham, NC: National Center for Child Traumatic Stress. Retrieved from <http://nctsn.org/products/caring-for-children-who-have-experienced-trauma>

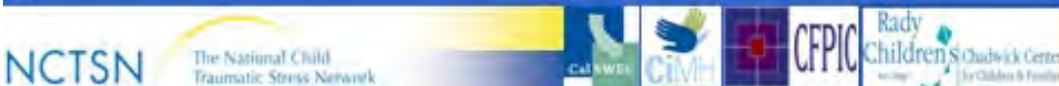


## Experience Grows the Brain *(continued)*

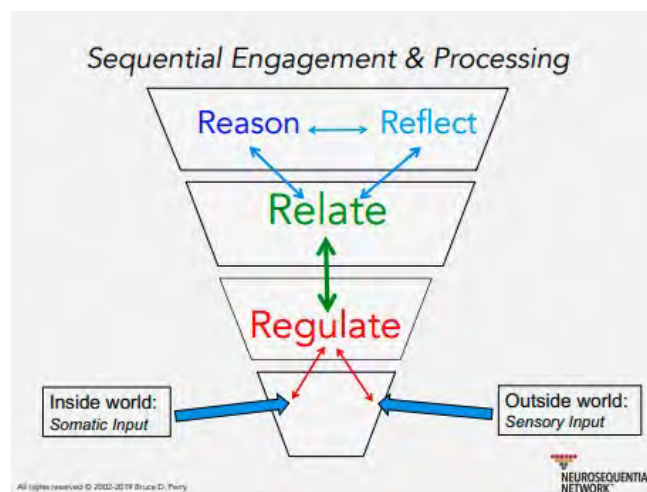
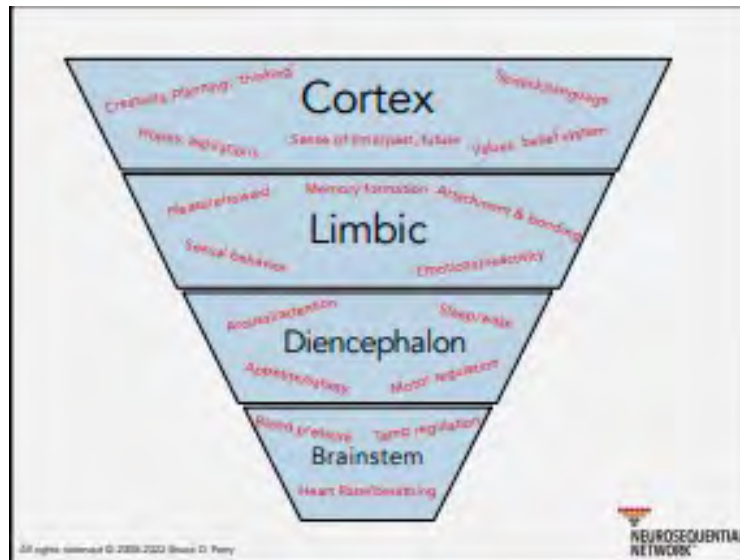
- The brain develops by forming connections.
- Interactions with caregivers are critical to brain development.
- The more an experience is repeated, the stronger the connections become.



Source: NCTSN: Caring for Children Who Have Experienced Trauma. Retrieved from <http://nctsn.org/products/caring-for-children-who-have-experienced-trauma>







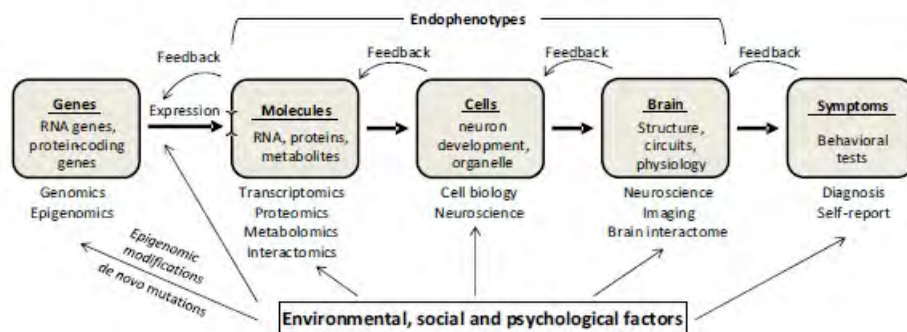


Sequential Neurodevelopment			
Age of most active growth	'sensitive' brain area	Critical functions being organised	Primary developmental goal
0-9 months	Brainstem	Regulation of arousal, sleep and fear states	State regulation, primary attachment, flexible stress response, resilience
6 months-2 years	Diencephalon	Integration of multiple sensory inputs	Sensory integration, motor control, relational flexibility, attunement
1-4 years	Limbic system	Emotional states, social language, interpretation of nonverbal information	Emotional regulation, empathy, affiliation, tolerance
3-6 years	Cortex	Abstract cognitive functions, socio-emotional integration	Abstract reasoning, creativity, respect, moral and spiritual foundations

## Literature Review – a trauma-sensitive approach for children aged 0-8 years

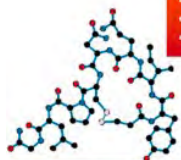
Funded by the Australian Government Department of Families, Community Services and Indigenous Affairs

422 Yihong Zhao and F. Xavier Castellanos J Child Psychol Psychiatr 2016; 57(3): 421-39




**Figure 1** A simplified flow chart for psychiatric disorders: from genes to symptoms. In this flow chart, results from one level (gray box) can exert feedback regulation at several levels upstream although only one level immediately upstream is shown for simplicity. Environmental impacts on each level are indicated. The studies for understanding each level and consequently the corresponding data types are listed below each level





**CRITICAL/SENSITIVE PERIODS**  
Neurobiological Mechanisms

- Change in balance of excitation to inhibition
- Involves activity at interneurons
- Increasing preference to selective environmental inputs
- Sequence of CPs from lower to higher brain functions
- Deprivation of essential inputs leads to brain reorganization



**OXYTOCIN**  
System Supporting SP Effects on Social Growth

- Organization of OT availability at critical limbic and neocortical sites depends on early caregiving
- OT directs young to preferentially select species specific social stimuli to form dyad-specific attachment
- OT receptors become connected to specific social cues via the system's experience-dependent plasticity
- Dendritic mode of OT release leads to feed-forward autoregulated functioning in response to experiences during SP

**BIOBEHAVIORAL SYNCHRONY**  
Experience Required during SP for Social Growth

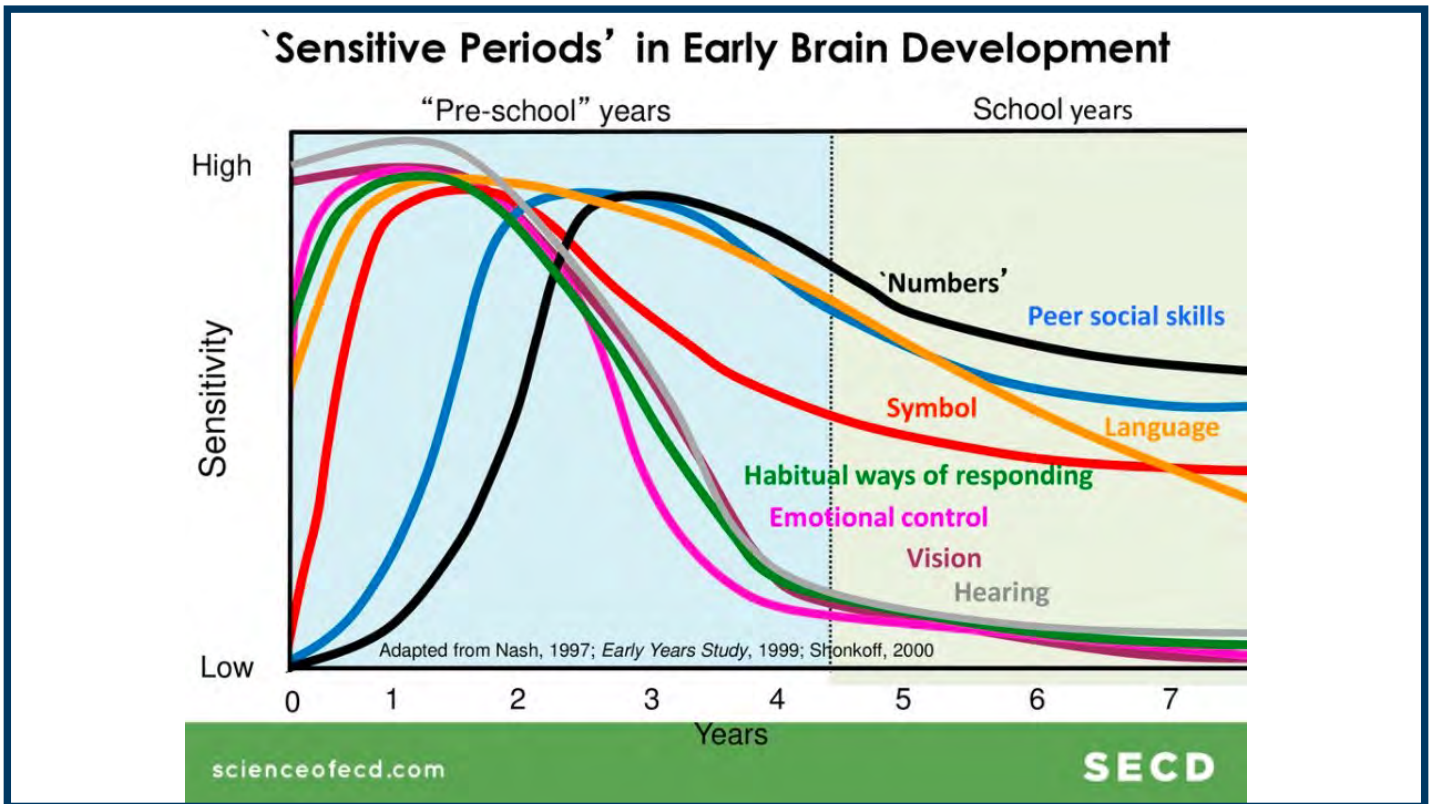
- Synchrony is the mechanism by which early environment exerts its effects via coordination of biological and social processes during social contact
- Biobehavioral synchrony in mammals occurs in the context of mother's body
- Human biobehavioral synchrony also includes the coordination of visuo-affective cues in the gaze, affect, vocal, and touch modalities
- Synchrony experienced during SP carries long-term effect on children's social growth, stress management, emotion regulation, and mental health

**Figure 2.** Critical periods (CP) involve specific neurobiological mechanisms observed across developmental domains (e.g., neuronal, physiological, sensory, motor). In the social domain, the oxytocin (OT) system employs these mechanisms to support social growth by utilizing processes of *biobehavioral synchrony*. The experience of synchrony during sensitive periods for social growth in turn shapes the infant's OT functionality across mammalian species. SP, Sensitive periods.

*Development and Psychopathology* 27 (2015), 969–995  
© Cambridge University Press 2015  
doi:10.1017/S0954579415000048

RUTH FELDMAN  
Bar-Ilan University

Sensitive periods in human social development: New insights from research on oxytocin, synchrony, and high-risk parenting





**Serve and return** interactions shape brain architecture. When an infant or young child babbles, gestures, or cries, and an adult responds appropriately with eye contact, words, or a hug, neural connections are built and strengthened in the child's brain that support the development of communication and social skills. Much like a lively game of tennis, volleyball, or Ping-Pong, this back-and-forth is both fun and capacity-building. When caregivers are sensitive and responsive to a young child's signals and needs, they provide an environment rich in serve and return experiences.





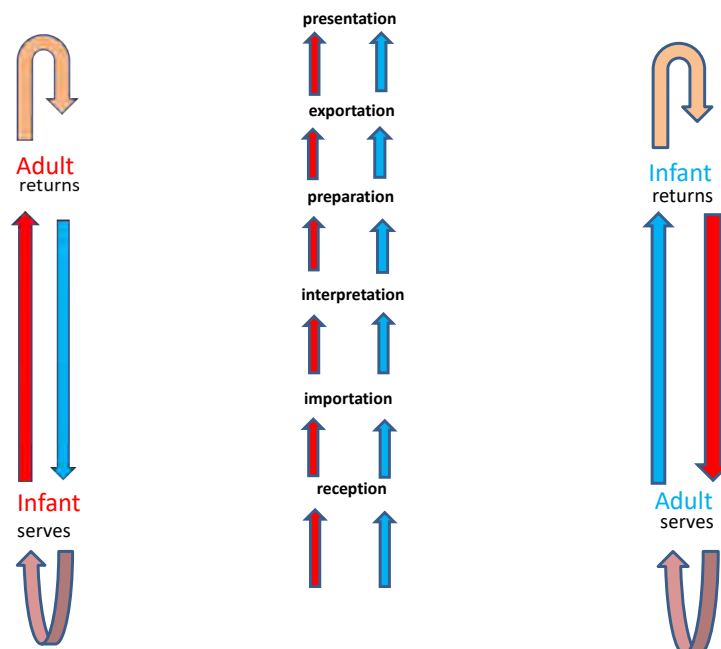


**Because responsive relationships are both expected and essential, their absence is a serious threat to a child's development and well-being.**

Healthy brain architecture depends on a sturdy foundation built by appropriate input from a child's senses and stable, responsive relationships with caring adults. If an adult's responses to a child are unreliable, inappropriate, or simply absent, the developing architecture of the brain may be disrupted, and subsequent physical, mental, and emotional health may be impaired. The persistent absence of serve and return interaction acts as a "double whammy" for healthy development: not only does the brain not receive the positive stimulation it needs, but the body's stress response is activated, flooding the developing brain with potentially harmful stress hormones.



 Center on the Developing Child  
HARVARD UNIVERSITY







## Optimal Early Caregiving

- Present
  - Quantity matters
- Attentive
  - To the infant/child
- Attuned
  - Accurately interpret non-verbal cues
- Responsive
  - Respond to the needs of the infant

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NEUROSEQUENTIAL  
NETWORK™

[Neuro Endocrinol Lett.](#) 2011;32(2):111-20.  
**Attachment in integrative neuroscientific perspective.**  
[Hruby B, Hasto J, Minarik P.](#)

*Radovan Hruby, Jozef Hasto, Peter Minarik*

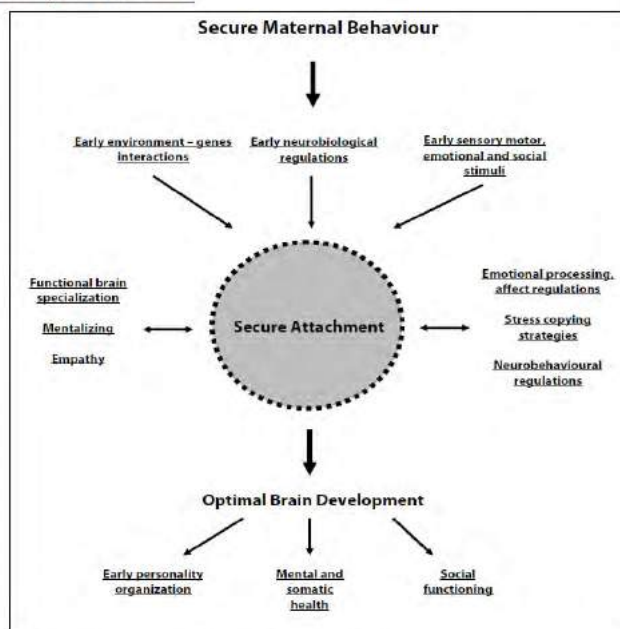
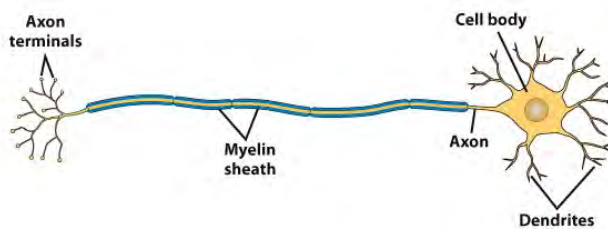
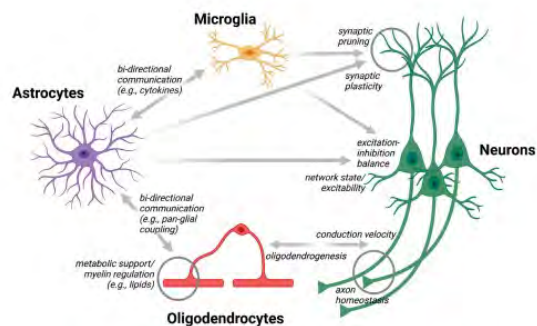


Fig. 1. Attachment and its importance for neurodevelopmental regulations.



The Alcohol Pharmacology Education Partnership is powered by WordPress at Duke WordPress Sites.

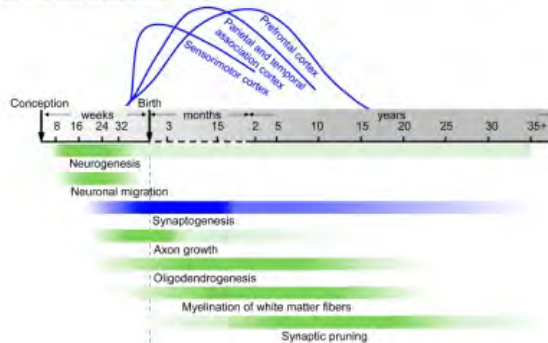


**FIGURE 3** Astrocytes regulate neural circuits directly and indirectly. Astrocytes can modulate neural circuit function directly through different mechanisms (see Figure 1). Microglia have also been shown to control neuronal synapse number and activity on various timescales. Similarly, oligodendrocytes can regulate neurons' axonal properties (e.g., conduction velocity) in an activity- and behavior-dependent manner. Astrocytes bidirectionally communicate with and regulate both microglia and oligodendrocytes (e.g., through diffusible messengers and physical interactions), allowing them to influence neural circuits indirectly and in a spatially and temporally distinct manner than their direct routes.

**How to cite this article:** Hirrlinger, J., & Nimmerjahn, A. (2022). A perspective on astrocyte regulation of neural circuit function and animal behavior. *Glia*, 70(8), 1554–1580. <https://doi.org/10.1002/glia.24168>



**Figure 1:** Timeline of spatiotemporally distinctive human brain maturational processes, including neurogenesis, neuronal migration, synaptogenesis, axon growth, oligodendrogenesis, myelination of white matter fibers and synaptic pruning. Time axis is in post-conceptual weeks (before birth), postnatal months (until 24 months), and postnatal years (after 2 years). The color intensity in each bar corresponds to the rate of developmental changes. The spatial progression across brain regions is illustrated using synaptogenesis (blue bar) as an example. Specifically, the spatial progression of synaptogenesis from primary sensorimotor cortex to higher-order prefrontal cortex is illustrated by the blue curves above the time axis.



Minhui Ouyang, Jessica Dubois, Qinlin Yu, Pratik Mukherjee, Hao Huang. Delineation of early brain development from fetuses to infants with diffusion MRI and beyond. *NeuroImage*, 2019, 185, pp.836-850. 10.1016/j.neuroimage.2018.04.017 . hal-02436254

**Major speech, language, and communication milestones\***

Age	Expressive language skill	Receptive language skill	Gestural communication skill
Birth to 2 months	Cries	Turns toward sound	-
2 to 4 months	Coos (makes open vowel sounds: "ooh"; "aah")	Social smile Watches faces intently	-
6 months	Babbles (repetitive consonant-vowel combinations: "bababa"; "mama")	Responds to name	-
12 months	Says first word/word approximation Uses jargon/babbles with inflection Repeats sounds or gestures to get attention	Follows 1-step verbal command with gesture (eg, "Give me the ball") Responds to "no" (eg, stops activity)	Visually follows adult's pointing Points to request (12 to 14 months) Starts to use gestures (eg, shaking head "no")
15 to 18 months	Points to common body parts when named	Follows 1-step verbal command without gesture	Points to share attention/enjoyment with another person (not just to request things) Shows objects to another person
18 to 24 months	Uses <sup>†</sup> 2-word phrases ("More milk"; "go outside")	Points to objects or people when named	-
24 to 36 months	Answers simple questions ("What's your name?"; "Who's that?") 50% intelligible	Follows 2-step verbal command	-
36 to 48 months	Uses 4- to 5-word sentences Uses pronouns and some plurals 75% intelligible	Understands placement in space (in, in, under)	-
48 to 72 months	Uses full sentences with grammatical markings (eg, plurals, verb endings) 100% intelligible	Follows 3-step verbal command	-

\* A general knowledge of skills that are typical for age (milestones/50<sup>th</sup> percentile skills) is important in order to conduct general developmental surveillance in primary care. However, milestones are not particularly helpful in making decisions about which children may require more formal evaluation or closer monitoring. This is because of the significant variability in the normal range of development of different skills in young children. Red flags (usually the 90<sup>th</sup> percentile for skill attainment) are more helpful in clinical decision-making<sup>11</sup>.

<sup>†</sup> Young children's use of words implies spontaneous expression of words or word approximation to communicate a request or to interact with another person. Echoing or immediately repeating words spoken by a caregiver does not constitute having or using words in the meaning of the milestone.

Reference:  
1. Sills L. Use of developmental milestones in pediatric residence training and practice. [Time to rethink the meaning of the mean. *J Dev Behav Pediatr* 2007; 28:47].



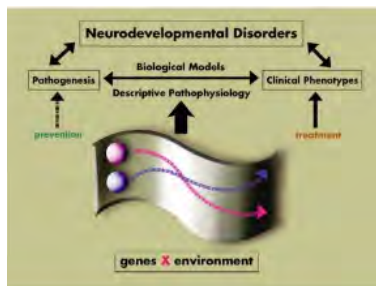
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**Table 1: The continuum of children's and infants' experiences of mental health**

Point on the continuum	General description	Infant-specific description
<b>Healthy</b>	Children experience a state of positive mental health and wellbeing.	Infants experience a state of positive mental health and wellbeing.
<b>Coping</b>	Children experience challenges to their mental health but are equipped with the mental resources to manage these effectively.	Infants may experience challenges but have the nurturing support and care to be able to help them cope with these challenges.
<b>Struggling</b>	Children experience challenges to their mental health, and but are not managing these effectively and need additional support.	If infants are struggling with their mental health, they and their families will need additional support.
<b>Unwell</b>	Children experience mental illness and considerable challenges to their wellbeing. They need additional support to manage and recover.	Mental illness is not usually diagnosed in infants. However, infants who show substantial signs of struggling will require extra support for themselves and their families.

National Mental Health Commission. (2021). Draft national children's mental health and wellbeing strategy. Canberra: National Mental Health Commission. Available [here](#).



**Figure 1. Clinical Disorders and Neural Development**

Differences between individuals in genetic vulnerability (red and blue circles) for a neurodevelopmental disorder confer, among other features, distinct responses to the environmental terrain, which together influence brain development. Gene-by-gene interactions directly influence developmental trajectory (colored lines), resulting in the expression of distinct phenotypes that are characteristic of a particular disorder. Biological models (e.g., genetic mutants, in vitro systems, protein-protein interactions) and descriptive pathophysiology (e.g., postmortem, genetics, imaging, behavioral/cognitive assessment) contribute to hypotheses regarding disease pathogenesis and to defining the phenotypes specific to a neurodevelopmental disorder. While evidence-based treatments are rooted in large part on successes in impacting a particular clinical phenotype (e.g., language, cognitive, social interventions), prevention of neurodevelopmental disorders will come from understanding the details of their origin (pathogenesis).

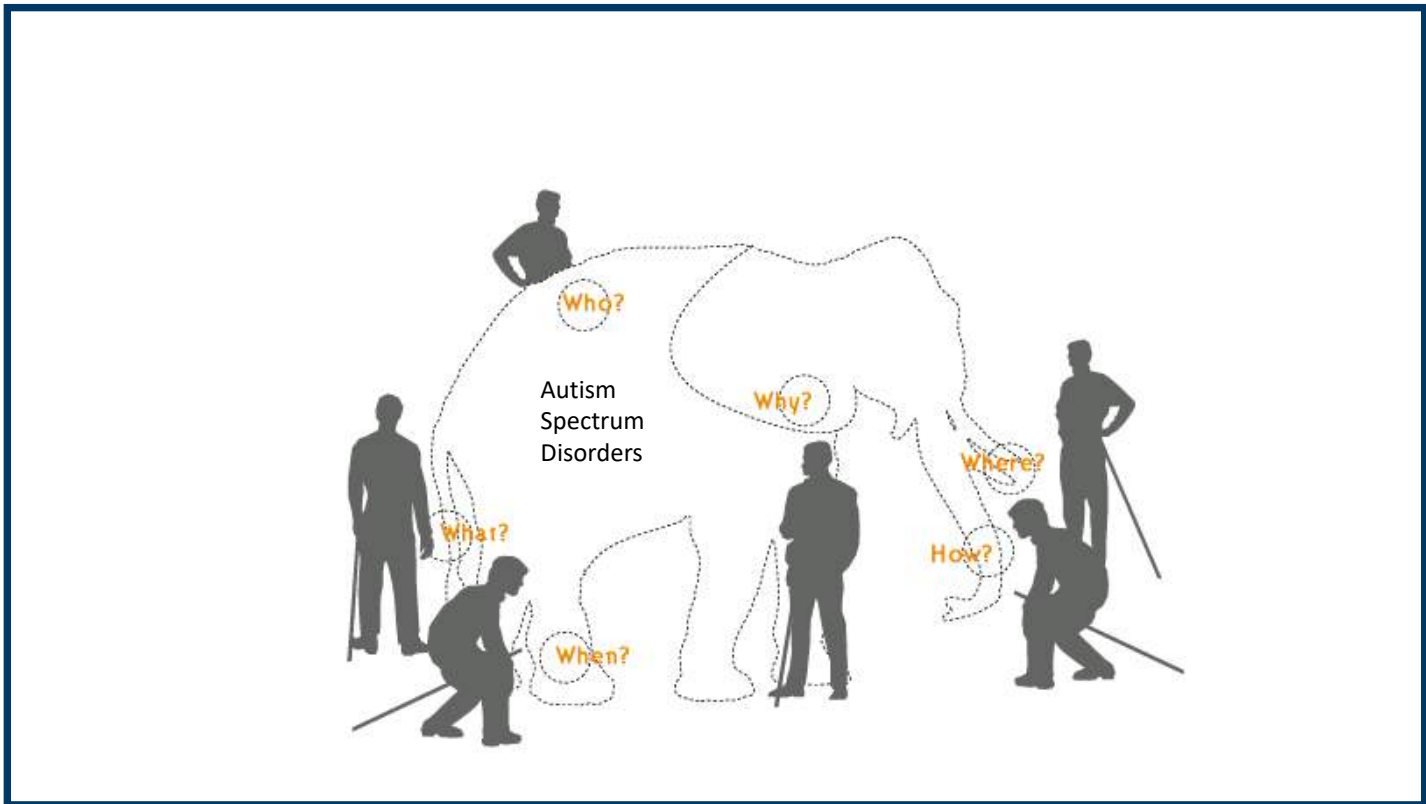
Neuron, Vol. 46, 407-412, May 5, 2006, Copyright ©2005 by Elsevier Inc. DOI 10.1016/j.neuron.2005.04.019

**Developmental Neurobiology and Clinical Disorders: Lost in Translation?**

Minireview

Pat Levitt\*

ment.\* This makes perfect sense to developmental



**Results:** For 2020, across all 11 ADDM sites, ASD prevalence per 1,000 children aged 8 years ranged from 23.1 in Maryland to 44.9 in California. The overall ASD prevalence was 27.6 per 1,000 (one in 36) children aged 8 years and was 3.8 times as prevalent among boys as among girls (43.0 versus 11.4). Overall, ASD prevalence was lower among non-Hispanic White children (24.3) and children of two or more races (22.9) than among non-Hispanic Black or African American (Black), Hispanic, and non-Hispanic Asian or Pacific Islander (A/PI) children (29.3, 31.6, and 33.4 respectively). ASD prevalence among non-Hispanic American Indian or Alaska Native (AI/AN) children (26.5) was similar to that of other racial and ethnic groups. ASD prevalence was associated with lower household income at three sites, with no association at the other sites.

**Suggested citation for this article:** Maenner MJ, Warren Z, Williams AR, et al. Prevalence and Characteristics of Autism Spectrum Disorder Among Children Aged 8 Years — Autism and Developmental Disabilities Monitoring Network, 11 Sites, United States, 2020. *MMWR Surveill Summ* 2023;72(No. SS-2):1–14. DOI: <http://dx.doi.org/10.15585/mmwr.ss7202a1>



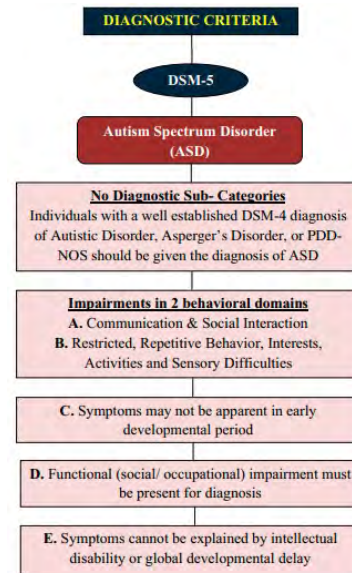


**Table 1** Criteria for diagnosis of Autism (DSM-5)

Social communication and interaction domain	Repetitive and restrictive behavior domain
<b>1. Deficits in social emotional reciprocity</b> Abnormality in social approach Failure of normal back and forth conversation Decreased sharing of interests, emotions, affect and response Total lack of initiation of social interaction	<b>1. Stereotyped or repetitive speech, motor movements, or use of objects</b> Simple motor stereotypes Echolalia Repetitive use of objects Idiosyncratic phrases
<b>2. Deficits in non-verbal communicative behaviors</b> Poorly integrated verbal and non-verbal communication Abnormal eye contact and body-language Difficulty in understanding and use of non-verbal communication Complete absence of facial expression or gestures	<b>2. Excessive adherence to routines, ritualized patterns of behavior</b> Excessive resistance to change such as motor ritual Insistence on same route or food Repetitive questioning or extreme distress at small changes
<b>3. Deficits in developing and maintaining relationships</b> Difficulty making friends Apparent absence of interest in people Difficulties adjusting behavior to suit different situations	<b>3. Highly limited, fixed interests which are abnormal in intensity or focus</b> Strong attachment to and/or preoccupation with strange objects Excessively limited or conservative interests
	<b>4. Hyper- or hypo- reactivity to sensory input</b> Unusual curiosity in sensory aspects of environment Apparent indifference to heat/ pain/cold Adverse response to particular sounds or textures Excessive smelling or touching of objects Fascination with lights or spinning objects

Priya Joon<sup>1</sup> · Anil Kumar<sup>1</sup> · Milind Parle<sup>2</sup>

Pharmacological Reports (2021) 73:1255–1264  
<https://doi.org/10.1007/s43440-021-00244-0>



- 1. Deficits in social interaction:** Children with autism may have difficulty in social interaction, a paucity of gestures to express interest, and a reduced ability to initiate or sustain a conversation with others. The lack of social and emotional reciprocity manifests itself in difficulty in understanding the perspective of another person and sharing emotions, interests, or affect. These deficits manifest as poor child–parent interaction, difficulty in establishing relationships with peers and responding to nonverbal cues.
- 2. Deficits in nonverbal communication:** Nonverbal communication such as eye contact, gestures, and facial expressions might be inappropriate, poorly applied, or absent. Lack of or fleeting eye contact may be an important clue to ASD. The child may be difficult to engage and may be lost in his/her world.
- 3. Speech and language impairments:** Parents complain that the child has a significant speech delay. In about a third of cases, there may be a history of regression in the language sector. Often, the child does not respond to his/her name unless there is a matter that is of interest to him/her and may be suspected of being deaf. They may ignore even very loud noises, or have a fascination for particular sounds; other sounds may cause extreme distress. At times, the child may display an abnormal use of words, or keep speaking words without any meaning. Many children may be mute or exhibit repetition of heard words and phrases. For older children with normal intelligence, concerns about impaired pragmatic and semantic language skills may be significant. Some of the older children may be quite verbose and talk about

- their circumscribed interests in a monotone with limited emotional inflection or regard for the listener's response.
- 4. Lack of joint attention:** This is one of the core deficits and is manifested as the inability to share a focus of interest with another person. Some examples are lack of pointing to share attention, or not sharing interests, and not able to sustain back and forth gaze.
  - 5. Impairments in play:** Generally, there is a lack of interest in play involving social interaction; a lack of pretend play and sharing. Children may have difficulty playing with toys in a constructive manner, and play may be repetitive and sensation-seeking. For example, children may repetitively spin the wheels of a toy car, line the blocks in a row, stare at objects in an unusual manner, or spend considerable time banging and licking toys.
  - 6. Abnormal movements and behaviors:** Children with ASD often have difficulty staying still, and they may keep wandering, moving round in circles, keep spinning toys or banging objects. Flapping of hands is often seen, especially when excited. Stereotypic movements are common in younger children with autism. Repetitive motor behaviors, nonfunctional routines, intense preoccupation with parts of an object, and self-injurious behaviors are reported by parents and can also be easily observed in the clinic.
  - 7. Strong preference for sameness:** Children with autism may have a strong preference for adherence to routines and/or ritualized behavior patterns. These behaviors may show up as a child having difficulty with transitions, strictly following rules, or repetitive questioning.
  - 8. Sensory issues:** Some children with ASD can have significant sensory issues manifesting either as hypersensitivity or hyposensitivity to a range of visual, auditory, and vestibular stimuli. Most children with ASD experience a combination of both over- and under-responsiveness to bright lights, certain sounds, smells, tastes, or textures. Children with ASD can experience a constant need for movement, an attraction towards bright colors and lights, hugging and intrusive touching of strangers, frequent mouthing of nonfood items, indifference to pain or temperatures, unusual avoidance of certain textures, and plugging of ears to avoid loud or unusual sounds.

Indian Journal of Pediatrics  
<https://doi.org/10.1007/s12098-022-04363-1>

REVIEW ARTICLE

**Early Diagnosis of Autism Spectrum Disorder: What the Pediatricians Should Know**

Pratibha Singh<sup>1,2</sup> · Prabhjot Malhi<sup>2</sup>



Table 1. Symptoms of autism spectrum disorder: real life examples of red flags and pink flags from expert clinicians.

Symptom Type	Red Flag	Pink Flag
<b>Restricted, Repetitive Patterns of Behavior, Interests or Activities</b> Restricted Interests and Play	Exhaustive and obsessive interest in highly specific, atypical topics. For example, dishwasher models, electric blanket controls, state license plates, WWII war planes, recites the Latin names of dinosaurs to strangers at the grocery store, carries doorknocker with them at all times or memorizes bus routes as a hobby.	Really likes to learn about and talk about certain niche topics. For example, Minecraft, Dinosaurs, Thomas the Train, Fire Night at Freddy's, US History, Aviation, My Little Pony or Psychology.
Repetitive Movements	Stereotyped pacing that wears a route into the carpet due to frequency, whole body spinning and/or rocking in conjunction with head banging when content or bored or to wind down or pink flag movements combined with associated visual regard.	Non-specific pacing, toe walking, head banging when upset or frustrated, shaking legs up and down, wringing hands, hand flapping (not uncommon in young children), subtle finger posturing while talking or completing tasks.
Sensory Seeking Behaviors	Licking sandpaper, cannot go for walks on rainy days because child lies face down in puddles to feel water on lips, repeated smelling of items with no odor (e.g., puzzle pieces), Lining up items -and looking along the line (lights down on the floor to look at objects at eye level), peeing out of corner of eyes (visual regard), Backing one's body into another to request frequent and intense squeezing.	Likes rolling down hills, rollercoasters, always wants to spin in tire swings or office chairs, loves water play, seeks out spicy or crunchy foods, seeks out mirrors or bright lights, prefers tight clothes, likes tight huggi-squeezers, heavy blankets or weighted vests, likes walking barefoot, likes to stroke or rub hair.
Sensory under sensitivity, over sensitivity (sensory avoidant behaviors)	Under responsive: Major injury occurred without display of pain or shaming with adult (burned hand on stove, broken toe, needed stitches when closed hand in car door).  Oversensitive: Avoids favorite places because cannot stand the hum of neon lights, extreme distress with daily noises these cannot occur in their presence (e.g., vacuum), repulsed by the smell of people who are eating mints or have recently bathed and smell of soap, since infancy has avoided or resisted physical contact (touch).	Under responsive: High pain tolerance for minor injuries (skinned knee, bruises).  Oversensitive: Picky eater, dislikes soft texture or mixed texture food, refuses hot or cold food (insists on room temperature), dislikes tags in clothes, hates having hair washed or cut, refuses to wear jeans, shoes, or jackets, resists change of clothes with change of seasons, Dislikes or is distressed by loud noises (fire alarm, siren), covers ears with blender, likes to be squeezed or tapped but not touched softly or stroked, Will initiate touch with others but dislikes others to initiate touch.
Difficulty with Transitions and Change, Rigidity or Inflexibility	Severe distress with trivial changes (e.g., home decor is moved, need to take alternate route due to roadwork), even switching from non-preferred to preferred activities is hard (e.g., Let's skip teeth brushing tonight and read an extra book instead), Refuses to eat from bowls, always walks on the left side of sidewalk.	Adjusting to new teachers or substitutes or returning to school after a holiday) is stressful, switching from preferred to non-preferred activities is hard (e.g., time to turn off TV and get ready for bed), has to complete activities (TV program, game, worksheet), Needs special loverly to fall asleep, preference for a certain seat in the car or favorite plate.

(continued)

Table 1. Continued.

Symptom Type	Red Flag	Pink Flag
Play, whole and part relationships	Little functional use of toys as they are intended to be used (e.g., exclusively spins wheels on cars but never "drive them"), interest in objects to the exclusion of people or the social world.	Poor quality pretend play (pretend play by him/herself but not with others, pretends same scenario over and over), wants others/caregiver to participate in play but only in certain ways (e.g., may be very directive).
<b>Social Communication and Social Interaction</b>		
Social Relationships	Seeks out relationships for primarily rational reasons (e.g., cites tax benefits of marriage). Talks incessantly about preferred topics regardless of partner's interest. Not easily comforted by caregiver, and distress may have no obvious cause.	Trouble understanding and expressing feelings or emotions (e.g., alexithymia), trouble reading the tone of a room, gravitates to adults or much younger children. May be difficult to comfort but caregivers usually know what the trigger for distress is. A history of difficulties maintaining friendships (often without understanding why they end).
Verbal Social Communication	Asks perseverative questions he/she already knows the answer to (not reassurance seeking), pronoun reversal (e.g., says "she wants water" instead of "I want water"), Immediate and delayed echolalia of content and tone (e.g., parroting repetitively without context, responds to "How are you?" with "Whenever you're in trouble, just yell for help") Fervent atypical prosody with combinations of ASD specific patterns (mid-word dysfluency/breathy breaks, poor intonation, mis-assigned stress) present since early childhood or marked language regression (loss of skill).	Scripted questions of others (asks new people same set of questions: What do you like to do? Did you have a nice weekend?), pedantic, overly formal speech (e.g., like a little professor), Immediate echolalia of content (e.g., responds to other's comment of "I like cows" with "cows", can be common in language delay), Subtle vocal quality differences or atypical prosody (e.g., tends to be flat, often exaggerated or frequent sarcastic tone), Speaks too loud or too soft for the social context, language delay with plateau of skills.
Nonverbal Social Communication	Using another person's hand as a tool (e.g., manipulates another's hand to operate a toy without eye contact), does not point to items just to show and share (e.g., point and look to airplane, then looks to parent with smile, then looks to airplane), regularly avoids eye contact and does not smile with eye contact to share enjoyment, even with preferred adults.	Leads others by the hand to what they want. Limited gestures, variable or poorly modulated eye contact. Does not respect the usual personal space boundaries. Has flat or inappropriate facial expressions.
Social Responsiveness, Social Initiation and Social Maintenance	Poor reciprocity (does not roll ball back and forth or respond to name when younger), never responds to comments made by others only direct questions, does not even notice if others are in obvious distress. Initiates with others solely to get needs met (e.g., requests). May tolerate (or enjoy) if caregiver or others join in child's play but child does not readily seek out the caregiver to share pleasurable activities or seek to maintain interaction if caregiver stops attending.	Trouble keeping a conversation going, only understands others' emotions if obviously displayed. Passive, abrasive, aggressive or disruptive when approaching another for social interaction. Described as being ignored by peers (due to passive presentation). Difficulty with reading nuances of peer relationships (e.g., is bullied OR reports being bullied even when that is not the intent; misunderstandings related to misinterpreting others' cues)

To cite this article: Susanne Duvall, Kira Armstrong, Ambreen Shahabuddin, Caroline Grant, Deborah Fein & Catherine Lord (2022) A road map for identifying autism spectrum disorder: Recognizing and evaluating characteristics that should raise red or "pink" flags to guide accurate differential diagnosis, *The Clinical Neurophysiologist*, 36(5), 1172-1207, DOI: 10.1080/13854656.2021.1921278

## Signs of autism spectrum disorders

A child with an ASD might:
Not point at objects to show interest (point at an airplane flying over)
Not look at objects when another person points at them
Have trouble relating to others or not have an interest in other people at all
Avoid eye contact and want to be alone
Have trouble understanding other people's feelings or talking about their feelings
Prefer not to be held or cuddled or might cuddle only when they want to
Appear to be unaware when other people talk to them but respond to other sounds
Be very interested in people, but not know how to talk to, play with, or relate to them
Repeat or echo words or phrases said to them, or repeat words or phrases in place of normal language (echolalia)
Have trouble expressing their needs using typical words or motions
Not play "pretend" games (pretend to feed a doll)
Repeat actions over and over again
Have trouble adapting when a routine changes
Have unusual reactions to the way things smell, taste, look, feel, or sound
Lose skills they once had (for instance, stop saying words they were once using)

Centers for Disease Control and Prevention, Autism spectrum disorder (ASD). Facts about ASD. Available at: [www.cdc.gov/ncbddd/autism/facts.html](http://www.cdc.gov/ncbddd/autism/facts.html).

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## Social Communication and Interaction Skills

Social communication and interaction skills can be challenging for people with ASD.

Examples of social communication and social interaction characteristics related to ASD can include

- Avoids or does not keep eye contact
- Does not respond to name by 9 months of age
- Does not show facial expressions like happy, sad, angry, and surprised by 9 months of age
- Does not play simple interactive games like pat-a-cake by 12 months of age
- Uses few or no gestures by 12 months of age (for example, does not wave goodbye)
- Does not share interests with others by 15 months of age (for example, shows you an object that they like)
- Does not point to show you something interesting by 18 months of age
- Does not notice when others are hurt or upset by 24 months of age
- Does not notice other children and join them in play by 36 months of age
- Does not pretend to be something else, like a teacher or superhero, during play by 48 months of age
- Does not sing, dance, or act for you by 60 months of age



 Centers for Disease Control and Prevention  
CDC 24/7: Saving Lives, Protecting People™

Autism Spectrum Disorder (ASD)

[ASD Homepage](#) - [ASD Diagnosis, Treatment, and Services](#)

## Restricted or Repetitive Behaviors or Interests

People with ASD have behaviors or interests that can seem unusual. These behaviors or interests set ASD apart from conditions defined by problems with social communication and interaction only.

Examples of restricted or repetitive behaviors and interests related to ASD can include

- Lines up toys or other objects and gets upset when order is changed
- Repeats words or phrases over and over (called echolalia)
- Plays with toys the same way every time
- Is focused on parts of objects (for example, wheels)
- Gets upset by minor changes
- Has obsessive interests
- Must follow certain routines
- Flaps hands, rocks body, or spins self in circles
- Has unusual reactions to the way things sound, smell, taste, look, or feel



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## Other Characteristics

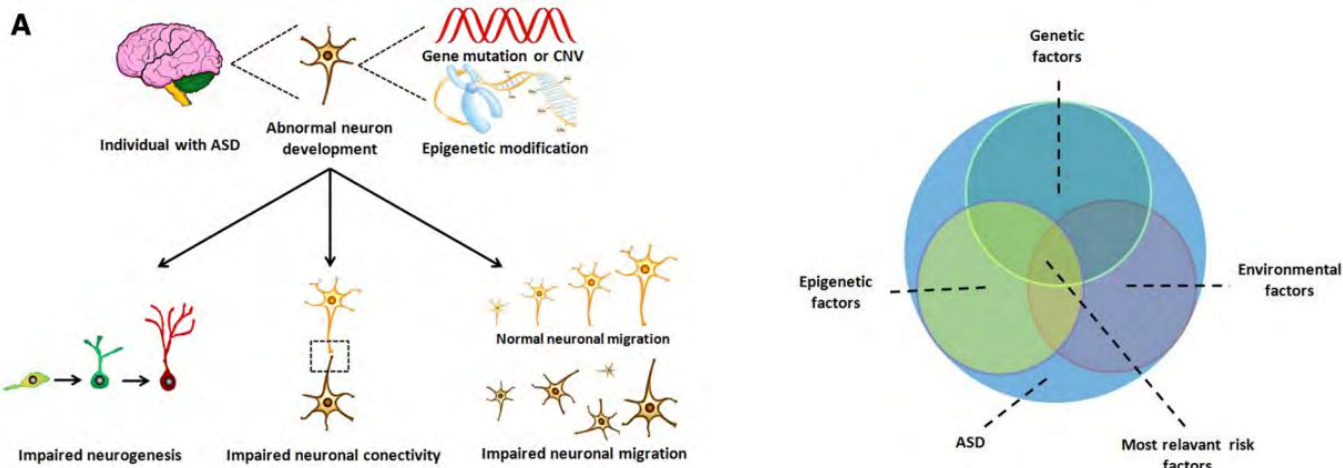
Most people with ASD have other related characteristics. These might include

- Delayed language skills
- Delayed movement skills
- Delayed cognitive or learning skills
- Hyperactive, impulsive, and/or inattentive behavior
- Epilepsy or seizure disorder
- Unusual eating and sleeping habits
- Gastrointestinal issues (for example, constipation)
- Unusual mood or emotional reactions
- Anxiety, stress, or excessive worry
- Lack of fear or more fear than expected



## Autism Spectrum Disorder (ASD)

[ASD Homepage](#) · [ASD Diagnosis, Treatment, and Services](#)



World Journal of Pediatrics (2019) 15:17–25  
<https://doi.org/10.1007/s12519-018-0210-2>

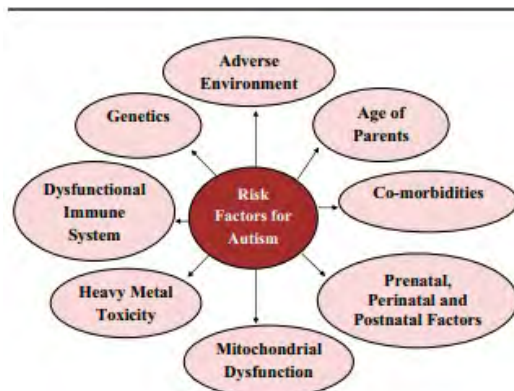


Fig. 1 Risk factors responsible for Autism

Priya Joon<sup>1</sup> · Anil Kumar<sup>1</sup> · Milind Parle<sup>2</sup>

Pharmacological Reports (2021) 73:1255–1264  
<https://doi.org/10.1007/s43440-021-00244-0>

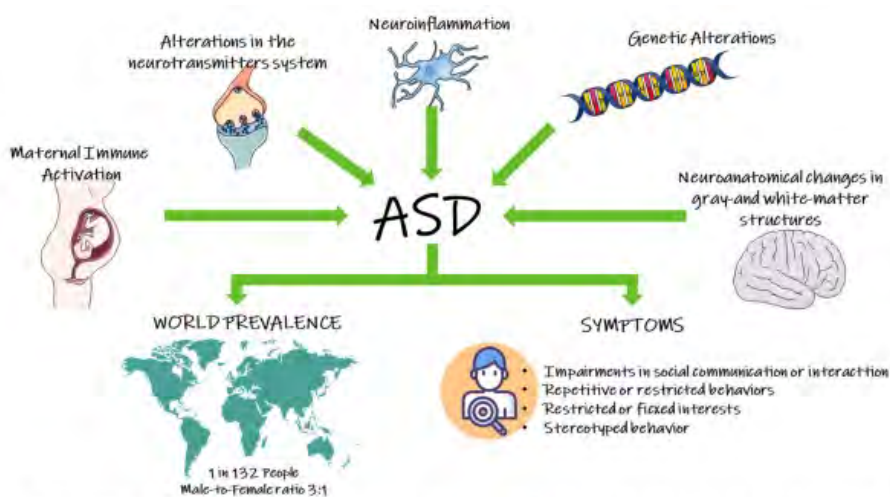


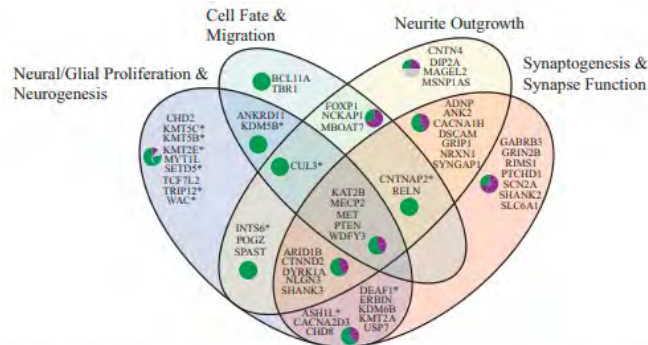
Fig. 1. Etiology and symptoms of autism spectrum disorders. The etiology and pathogenesis of ASD have not been completely identified. However, the combination of genetic and environmental factors and immune dysfunction can be underlying ASD development.

#### Neuroinflammation in autism spectrum disorders: Exercise as a “pharmacological” tool

Christiane V.A. Toscano<sup>a,1</sup>, Leonardo Barros<sup>b,1</sup>, Ahlan B. Lima<sup>c</sup>, Thiago Nunes<sup>d</sup>, Humberto M. Carvalho<sup>c</sup>, Joana M. Gaspar<sup>d,e,g</sup>

Neuroscience and Biobehavioral Reviews 129 (2021) 63–74





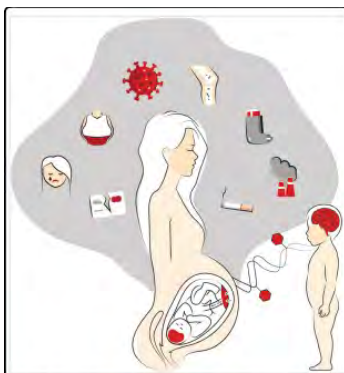
**Fig. 5** Distribution of 58 *hcASD* genes in four main categories of neural development. Functional annotation could be found for 58 *hcASD* genes based on a manual literature search. Most highly penetrant ASD genes are pleiotropic, being involved in multiple stages of brain development. The small pie-charts in each region indicate the percentage of genes in green and purple clusters from Fig. 4. The gray color in pie-charts represents percentage of genes with no strong expression level in fetal and early post-natal periods. A gene is marked by an asterisk (\*) if its function was inferred from evidence in adult neural stem cells, embryonic or hematopoietic stem cells, central nervous system other than brain, or, for one gene, cancer (see Table S2 for details and references).

Molecular Psychiatry (2019) 24:88–107  
<https://doi.org/10.1038/s41380-018-0056-y>

**EXPERT REVIEW**

**The ASD Living Biology: from cell proliferation to clinical phenotype**

Eric Courchesne<sup>1</sup> · Tiziano Pramparo<sup>1</sup> · Vahid H. Gazestani<sup>1,2</sup> · Michael V. Lombardo<sup>3,4</sup> · Karen Pierce<sup>1</sup> · Nathan E. Lewis<sup>2,5,6</sup>



**Fig. 1** Maternal immune activation, triggered by acute and systemic chronic inflammation, is proposed to affect fetal neurodevelopment, through inflammatory and epigenetic mechanisms. Common maternal disease and environmental factors including obesity, gestational diabetes, pre-eclampsia, smoking, pollution, low socioeconomic status, depression, psychosocial stress, autoimmune diseases and asthma are implicated in systemic chronic inflammation. In addition, infection is involved in acute inflammation. These maternal inflammatory states play a key role in immune activation during pregnancy through the placenta and immature blood-brain barrier to cause dysfunction in the developing fetal brain and prime the child to be susceptible to future hits through microglia activation and epigenetic alterations, manifesting a spectrum of diverse neurodevelopmental outcomes with varied expression and progression.

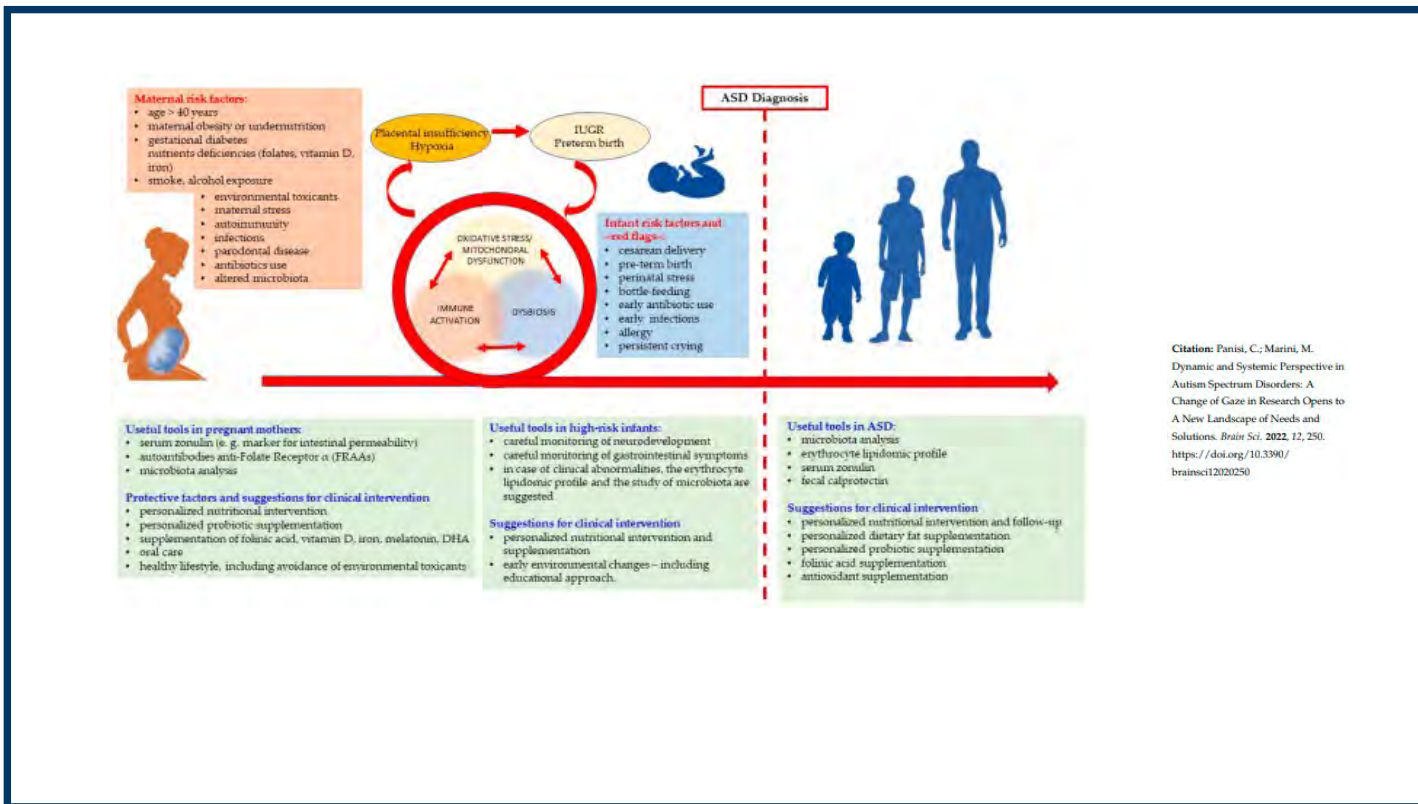
Han et al. *Translational Psychiatry* (2021)11:71  
<https://doi.org/10.1038/s41398-021-01198-w>

Translational Psychiatry

**ARTICLE**

**Open Access**

**Maternal acute and chronic inflammation in pregnancy is associated with common neurodevelopmental disorders: a systematic review**



Human development	Abnormalities induced by MIA and associated with ASD
1 <sup>st</sup> -2 <sup>nd</sup> Trimester	Upregulated cell cycle and downregulated migration and neurite outgrowth gene expression
	Cortical layering: over-production of neurons, increased cortical thickness, focal cortical dysplasia Cerebellar vermis dysplasia
2nd 3 <sup>rd</sup> Trimester	Microglia: enhanced priming, activation
	Dendritic morphology abnormalities GABAergic signaling, excitatory/inhibitory imbalance, number of interneurons
3 <sup>rd</sup> Trimester - postnatal	White matter neuron density
	Dendritic spines number and turnover rates Synaptic pruning and proteins
Postnatal	Early brain overgrowth
	Myelin functionality and stability
	Dopamine system
	Serotonin levels
	ASD-like abnormal social, vocalization, and ritualistic behaviors Gender-dependent effects Transgenerational effects

**The ASD Living Biology: from cell proliferation to clinical phenotype**

Article in *Molecular Psychiatry* · June 2018  
DOI: 10.1038/s41380-018-0004-4

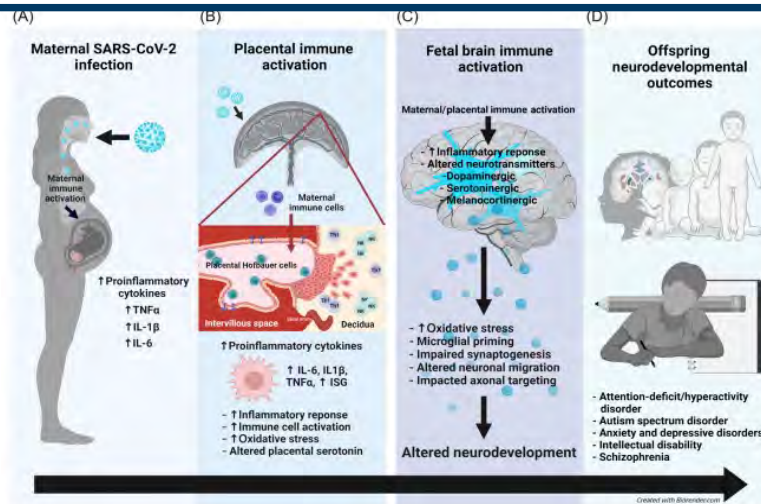


Figure 1. Coronavirus disease 2019 (COVID-19) in pregnancy and implications for offspring neurodevelopment. (A) Maternal severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) infection results in maternal immune activation (MIA) and increased proinflammatory cytokines in the maternal periphery. (B) Maternal SARS-CoV-2 infection can impact the placenta via two mechanisms: (i) placental immune activation and inflammation resulting from MIA (likely most common) or (ii) direct placental infection with SARS-CoV-2 (rare per the current literature). Placental immune activation and inflammation are associated with placental Hofbauer cell activation or priming, increased natural killer (NK) and T-helper 1 (Th1) cells at the maternal-fetal interface, increased proinflammatory cytokine production, upregulation of interferon-stimulated genes (ISGs), placental serotonin dysregulation, and increased oxidative stress. When direct placental infection with SARS-CoV-2 occurs (which is rare with the ancestral and other pre-Delta strains), the syncytiotrophoblast and cytotrophoblast layers are most commonly infected, and once the virus gains access to the intervillous space, it can theoretically gain access to the fetal circulation. (C) Both MIA and placental immune activation can lead to fetal brain immune activation, inflammation, and altered neurotransmitter signaling, including the serotonergic, dopaminergic, melanocortinergic, GABAergic, and glutamatergic systems. Fetal brain immune responses are associated with microglial priming, altered neural progenitor cell proliferation, impaired neuronal migration, synaptogenesis, and axonal targeting, all of which can result in altered offspring neurodevelopment. (D) Offspring affected by MIA are at increased risk for attention hyperactivity deficit disorder, autism spectrum disorder, anxiety, depression, impaired cognition, learning disabilities, and schizophrenia.

322 Trends in Molecular Medicine, April 2022, Vol. 28, No. 4

### COVID-19 in pregnancy: implications for fetal brain development

Lydia L. Shook,<sup>1,2</sup> Elinor L. Sullivan,<sup>3,4,5</sup> Janine O. Lo,<sup>6,7,8</sup> Roy H. Peris,<sup>9,10</sup> and Andrea G. Edlow<sup>1,2\*</sup>

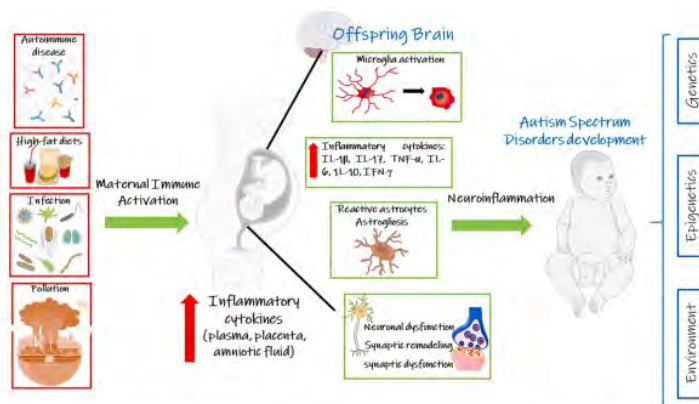


Fig. 2. Neuroinflammation associated with autism spectrum disorders. Maternal immune system activation is a risk factor that increases the chances of a child develops ASD. The increased inflammatory cytokines and autoantibodies that react to fetal brain tissue may change proper synaptic development in the offspring and that are linked to behavioral abnormalities seen in ASD, including repetitive behaviors, stereotypies, anxiety, and impaired social behaviors.

### Neuroinflammation in autism spectrum disorders: Exercise as a “pharmacological” tool

Christiane V.A. Toscano<sup>a,1</sup>, Leonardo Barros<sup>b,1</sup>, Ahlan B. Lima<sup>c</sup>, Thiago Nunes<sup>d</sup>, Humberto M. Carvalho<sup>e</sup>, Joana M. Gaspar<sup>a,4,\*</sup>

Neuroscience and Biobehavioral Reviews 129 (2021) 63–74





**Fig. 1 Potential mechanisms underlying the effects of maternal obesity on offspring neurodevelopment.** Maternal obesity alters both intrauterine environment and postnatal care through the promotion of oxidative stress, inflammation, lipotoxicity, and changes to the composition of breastmilk, all of which have detrimental effects on offspring brain health. Changes to fetal epigenetics, neural functioning,

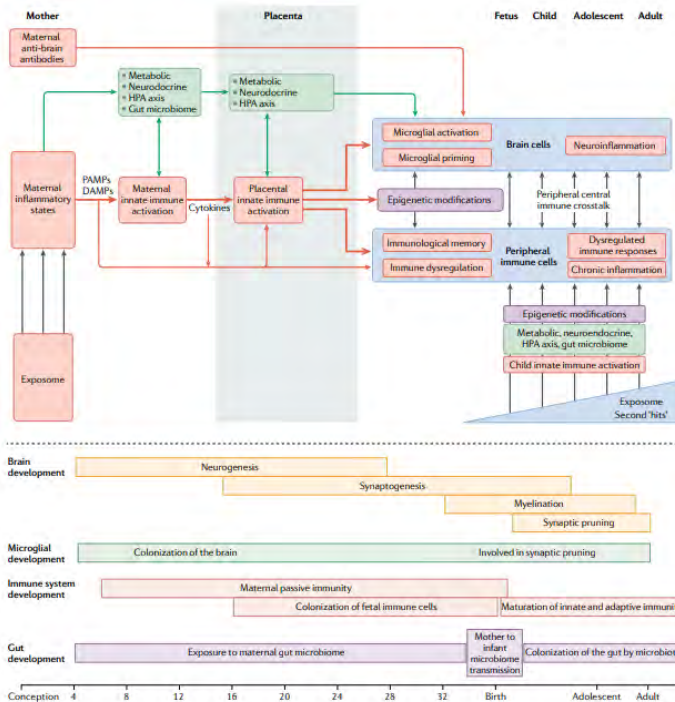
and microbiome due to maternal obesity also has detrimental effects of fetal neurodevelopment and can give rise of neurodevelopmental and neuropsychiatric disorders. (IL-1 interleukin-1, TNF- $\alpha$  tumor necrosis factor alpha, IL-6 interleukin-6, DHA docosahexaenoic acid, BDNF brain-derived neurotrophic factor).

Journal of Perinatology (2021) 41:928–939  
<https://doi.org/10.1038/s41372-020-00871-0>

REVIEW ARTICLE

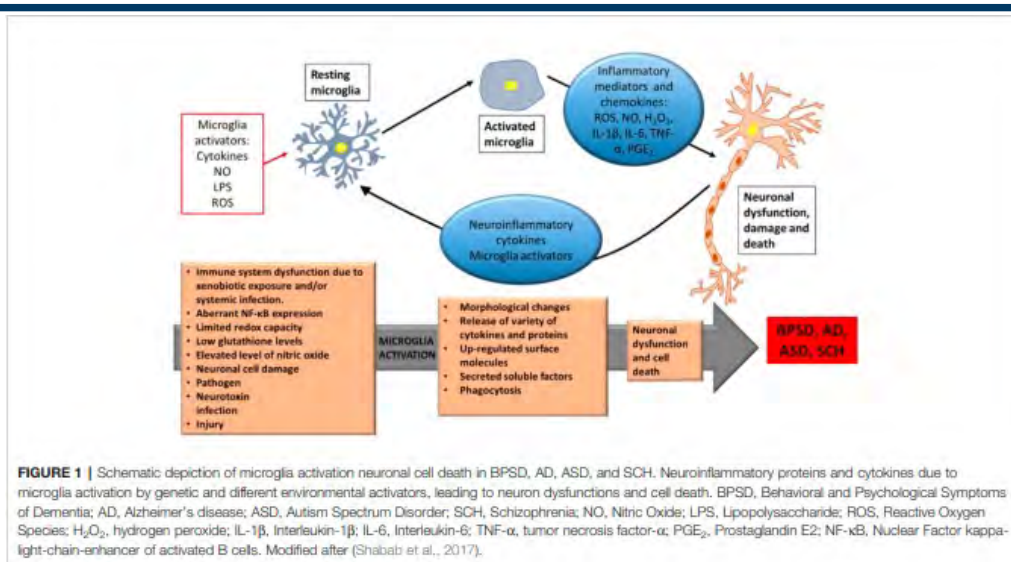
The impact of maternal obesity on childhood neurodevelopment

Lilin Tong<sup>1</sup> · Brian T. Kalish<sup>2</sup>



**Fig. 1 | Maternal immune activation and offspring development.** Evidence from human and animal studies indicates that maternal immune activation programmes the fetal brain and immune system through inflammatory and epigenetic mechanisms during key periods of CNS, microglial and immune system development, and colonization of gut microbiota. Heterogeneous infectious and non-infectious maternal inflammatory factors induce the release of pathogen-associated molecular patterns (PAMPs) and damage-associated molecular patterns (DAMPs), which activate Toll-like receptors on maternal peripheral innate immune cells and placental cells, leading to cytokine production<sup>12,14,15</sup>. Across the placenta, passive transport and active placental production of immune mediators occur and interact with transplacental metabolic, neuroendocrine and stress (hypothalamic–pituitary–adrenal (HPA))

signalling pathways<sup>16</sup>. The effects of maternal inflammation are proposed to induce long-lasting epigenetic memory on fetal microglia and immune cells during critical developmental periods<sup>17,18</sup>. The lower part of the figure shows the timings of key developmental processes in the offspring brain, immune system and gut microbiome. Postnatally, dynamic peripheral–central immune crosstalk occurs, involving peripheral inflammatory signals triggered by environmental immune-modifying factors and brain immune cells<sup>19</sup>. Interactions among offspring aberrant immune programming, genetic risk, sex and second immune ‘hits’ in life result in a state of chronic inflammation in both the brain and periphery, which manifest as lifelong neurobehavioural abnormalities<sup>7,15,21,19</sup>. Adapted from REF<sup>19</sup>, CC BY 4.0 (<https://creativecommons.org/licenses/by/4.0/>).

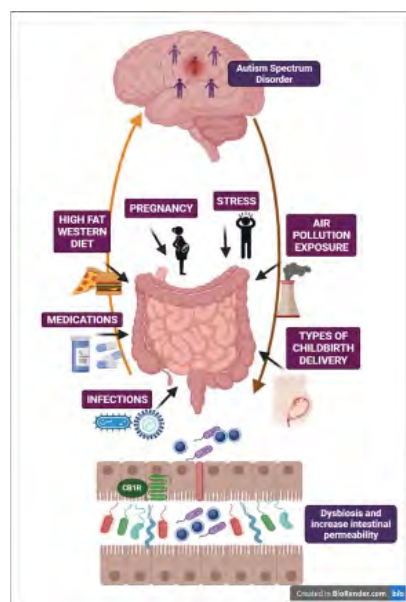


## Role of Neuroinflammation in Autism Spectrum Disorder and the Emergence of Brain Histaminergic System. Lessons Also for BPSD?

Nermin Eissa<sup>1,2</sup>, Adel Sadeq<sup>2</sup>, Astrid Sasse<sup>3</sup> and Bassem Stadek<sup>1,2\*</sup>

frontiers  
in Pharmacology

REVIEW  
published: 16 June 2020  
doi: 10.3389/fphar.2020.00885



**FIGURE 2** | The figure illustrates some critical mechanisms involved in derangement of gut microbiota ecosystem as potential threats increasing susceptibility to ASD. Gut microbiota and brain are reciprocally interconnected via multiple neural, endocrine and immune bidirectional pathways. Here are schematized brain-gut efferent pathways (e.g., HPA axis, autonomic nervous system) and gut-to-brain afferent pathways (e.g., vagal innervation and enteroendocrine signaling). Changes in gut microbiota composition and dysbiosis can be attributed to multiple threats, such as deleterious obesogenic diets (Western diet/high-fat diet), psychosocial and physical stressors, chronic medications due to medical co-morbidities, infections of multiple origins and exposure to air pollution. These environmental challenges may be viewed as pathogenic factors not only after birth, during postnatal neurodevelopment, but also during maternal gestation with relevance for infections occurring during pregnancy and the different types of childbirth delivery. Under the pressure of multiple threats, abnormal gut function and dysbiosis with pathobiont overgrowth may become the prevalent inflammatory condition, thus altering brain-gut axis reciprocal signaling and brain response. In turn, progressive loss of intestinal barrier function and consequent leaky gut increases the transfer of detrimental bacterial components to systemic circulation, intensifying chronic inflammation and susceptibility to ASD. The figure also depicts CB<sub>1</sub> receptors located at the intestinal epithelium, in proximity with neuroendocrine cells, which imply the role of eCB machinery in the regulation of gut barrier integrity.

## The Endocannabinoids-Microbiota Partnership in Gut-Brain Axis Homeostasis: Implications for Autism Spectrum Disorders

Roberto Coccarolo<sup>1,2\*</sup>, Maria Cristina Meronis<sup>3</sup> and Mauro Maccarrone<sup>1,2\*</sup>

frontiers | Frontiers in Pharmacology

REVIEW  
published: 03 July 2022  
doi: 10.3389/fphar.2022.898806



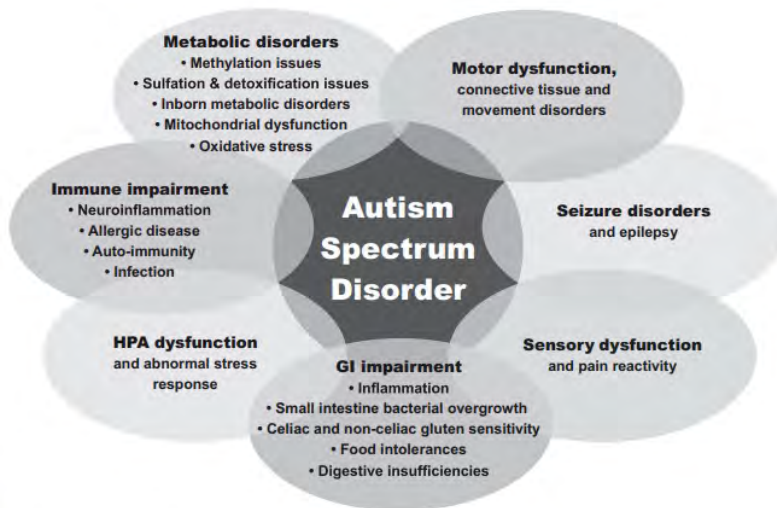


Figure 1 Physical health comorbidities associated with ASD.

Neuropsychiatric Disease and Treatment

Dovepress

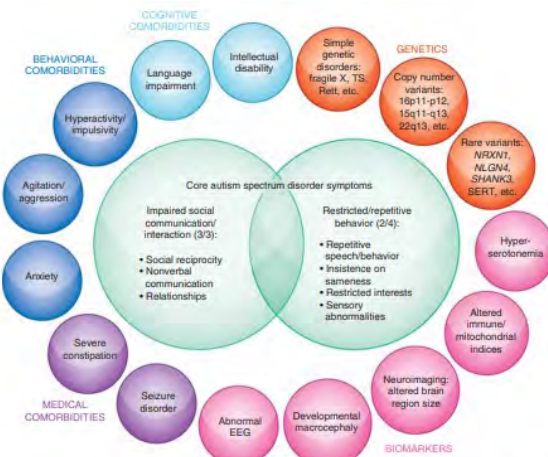
Open access to scientific and medical research

8 Peer Review on File

REVIEW

## Bridging the Gap Between Physical Health and Autism Spectrum Disorder

Regina Sala<sup>1</sup>  
 Lorene Amet<sup>2</sup>  
 Natasa Blagojevic-Stokic<sup>3</sup>  
 Paul Shattock<sup>4</sup>  
 Paul Whiteley<sup>5</sup>



Neuropsychopharmacology (2018) 37, 196–212  
 © 2018 American College of Neuropsychopharmacology. All rights reserved 0893-1200/18

REVIEW

## Networking in Autism: Leveraging Genetic, Biomarker and Model System Findings in the Search for New Treatments

Jeremy Veenstra-VanderWeele<sup>1,3,4</sup> and Randy D Blakely<sup>1,3,4</sup>

Figure 1. ASD symptoms, comorbidities, and biomarkers. The core symptoms of ASD are represented in the center and represent the common features required to receive a diagnosis. All three social communication/social interaction symptoms are required to receive a diagnosis in the DSM-V draft criteria. This domain represents the 'negative symptoms' of ASD, that is, absence of appropriate social communication. Two of the four restricted/repetitive behavior symptoms are required to receive a diagnosis in the DSM-V draft criteria. This domain represents the 'positive symptoms' of ASD, that is, the presence of unusual restricted, repetitive, or sensory behaviors. Around the periphery of the figure are symptoms or biomarkers that are not required for an ASD diagnosis but are more common in ASD than in the general populations. Quite a number of comorbid disorders or symptoms are seen in a substantial minority or even a majority of individuals with ASD, spanning cognitive, behavioral/psychiatric, and medical domains. As might be expected from the range of comorbid symptoms, biomarkers and genetic findings also reveal significant heterogeneity across individuals with ASD.

Neuropsychopharmacology **REVIEWS**



Thank you!



# Medical evaluation of Autism

Dr Moshe Kupferstein  
LEND/Healthfirst  
May 5, 2023

## Disclosures

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None



## About Me:

---

- Prior to medical school, I worked at a group home for developmentally disabled/medically frail children
- Pediatrician at outpatient practice-multispecialty office
- Completed mental health mini fellowship at the Reach Institute

[Patient-Centered Mental Health in Pediatric Primary Care \(PPP\)](#)—

## Presentation flow

---

- Role of pediatric providers during the well visit
- Screening tools
- Developmental milestones
- Definition of autism
- Definition of what's not autism
- Physical exam clues
- labs/imaging/genetics
- Take home points



## Well visit role of the pediatric provider

---

- Assess development at each well visit (surveillance)
  - Think developmental milestones in chart template
- Screen for developmental disabilities at 9,18,30
- Is the child growing and developing as expected for age?

## Diagnosis and treatment of autism in one slide

---

['Groundbreaking' autism test detects disorder in a single strand of hair](#)

[Scientists 'switch off' autism using \\$3 epilepsy drug: study](#)





## CDC Developmental milestones update-2022

---

Changed to 75%ile of children for that age

- Milestones are included at the age most ( $\geq 75\%$ ) children would be expected to demonstrate the milestone
- **Children's development did not change**, but the CDC's recommendations on when to worry or do something did.
- Eliminate "warning signs"
- Are able to be answered with yes, not yet, or not sure
- Use plain language, avoiding vague terms like may, can, and begins
- Are organized in developmental domains
- **Show progression of skills with age, when possible**
- Milestones are not repeated across checklists

As with all guidelines, there are some disagreement on this, notably from speech language pathologists

## Development domains

---

- Cognition
- Social
- Gross motor
- Fine motor
- Speech



## Social milestones

---

- 6 months
  - Knows familiar faces
  - Likes to look himself in the mirror
  - Laughs
- 9 months
  - Shy and clingy , fearful around strangers
  - Shows several facial expressions [ happy, sad, angry, surprised]
  - Looks when you call your name
  - Reacts when you leave [looks, reaches for you, cries]
  - Smiles or laughs when playing peek a boo

## Social milestones

---

- 12 months
  - Plays games with you, like pat-a-cake
- 15 months
  - Copies other children when playing with them
  - ***Shows you an object that he likes***
  - Claps when excited
  - Hugs stuffed doll or other toy
  - Shows affection [ hugs, kisses, cuddles]



## Social milestones

---

- 18 months
  - Puts hand out when you wash them
  - Looks at a few pages in a book with you
  - Helps you with getting dressed
- 24 months
  - Looks at your face to see how you react to new situations
  - *Notices when others are hurt*

## Tools for developmental screening

---

**Ages and stages (ASQ)**

**MCHAT**

Others:

PEDS

Denver development



## Ages and Stages

- Assess development over the five domains
- Takes about 10 minutes for parent to fill out
- About 2 minutes to score
- Paper or online versions available
- Parents can fill out in the waiting room
- [ASQ 9 month](#) ages and stages 9 months
- [ASQ 48 month sample](#) ages and stages 48 month sample
- CPT code : 96110



### 9 Month ASQ-3 Information Summary

9 months 0 days through  
9 months 30 days

Baby's name: \_\_\_\_\_ Date ASQ completed: \_\_\_\_\_

Baby's ID #: \_\_\_\_\_ Date of birth: \_\_\_\_\_

Administering program/provider: \_\_\_\_\_ Was age adjusted for prematurity when selecting questionnaire?  Yes  No

1. **SCORE AND TRANSFER TOTALS TO CHART BELOW:** See ASQ-3 User's Guide for details, including how to adjust scores if item responses are missing. Score each item (YES = 10, SOMETIMES = 5, NOT YET = 0). Add item scores, and record each area total. In the chart below, transfer the total scores, and fill in the circles corresponding with the total scores.

Area	Cutoff	Total Score	0	5	10	15	20	25	30	35	40	45	50	55	60
Communication	13.97		●	●	●	●	○	○	○	○	○	○	○	○	○
Gross Motor	17.82		●	●	●	●	●	○	○	○	○	○	○	○	○
Fine Motor	31.32		●	●	●	●	●	●	○	○	○	○	○	○	○
Problem Solving	28.72		●	●	●	●	●	●	○	○	○	○	○	○	○
Personal-Social	18.91		●	●	●	●	○	○	○	○	○	○	○	○	○



## 48 Month ASQ-3 Information Summary 45 months 0 days through 50 months 30 days

Child's name: John X. Smith Date ASQ completed: 11/18/2008  
 Child's ID #: 00123456789000000 Date of birth: 11/12/2004  
 Administering program/provider: Anytown Preschool/Ms. Jenkins

1. **SCORE AND TRANSFER TOTALS TO CHART BELOW:** See ASQ-3 User's Guide for details, including how to adjust scores if item responses are missing. Score each item (YES = 10, SOMETIMES = 5, NOT YET = 0). Add item scores, and record each area total. In the chart below, transfer the total scores, and fill in the circles corresponding with the total scores.

Area	Cutoff	Total Score	0	5	10	15	20	25	30	35	40	45	50	55	60
Communication	30.72	<b>25</b>	●	●	●	●	●	●	●	○	○	○	○	○	○
Gross Motor	32.78	<b>60</b>	●	●	●	●	●	●	●	●	●	●	●	●	●
Fine Motor	15.81	<b>20</b>	●	●	●	●	●	○	○	○	○	○	○	○	○
Problem Solving	31.30	<b>20</b>	●	●	●	●	●	○	○	○	○	○	○	○	○
Personal-Social	26.60	<b>60</b>	●	●	●	●	●	●	●	●	●	●	●	●	●

2. **TRANSFER OVERALL RESPONSES:** Bolded uppercase responses require follow-up. See ASQ-3 User's Guide, Chapter 6.

- |  |   |   |   |
|--|---|---|---|
| 1. Hears well?<br>Comments: <b>Ear infex, ear tubes, didn't talk until 2-3 yrs.</b>                              | Yes <input type="radio"/> NO <input checked="" type="radio"/> | 6. Family history of hearing impairment?<br>Comments:   | YES <input type="radio"/> No <input checked="" type="radio"/> |
| 2. Talks like other toddlers his age?<br>Comments: <b>Sentences and comperly not as advanced as younger kids</b> | Yes <input type="radio"/> NO <input checked="" type="radio"/> | 7. Concerns about vision?<br>Comments:  | YES <input type="radio"/> No <input checked="" type="radio"/> |
| 3. Understand most of what your child says?<br>Comments:   | Yes <input checked="" type="radio"/> NO <input type="radio"/> | 8. Any medical problems?<br>Comments: <b>Ear infex</b>  | YES <input checked="" type="radio"/> No <input type="radio"/> |
| 4. Others understand most of what your child says?<br>Comments:  | Yes <input type="radio"/> NO <input checked="" type="radio"/> | 9. Concerns about behavior?<br>Comments:  | YES <input type="radio"/> No <input checked="" type="radio"/> |
| 5. Walks, runs, and climbs like other toddlers?<br>Comments:   | Yes <input checked="" type="radio"/> NO <input type="radio"/> | 10. Other concerns?<br>Comments: <b>Language level- doesn't recognize numbers or letters yet.</b> | YES <input checked="" type="radio"/> No <input type="radio"/> |



	85%	DENVER II: 88%	IDI: 77% CDI: 88%	70%-80%	93.6% (POSI 6-30 months) 75% (POSI 31-48 months) 77% (SWYC Milestones) Above 70% (PPSC) 40.8% (POSI 6-30 months) 47.8% (POSI 31-48 months)
<b>Reading Level</b>	4th-6th grade level	Unknown	7th-8th grade level	PEDS: 4 <sup>th</sup> -5 <sup>th</sup> grade reading level DM: 1 <sup>st</sup> -2 <sup>nd</sup> grade reading level	Unknown
<b>Initial cost</b>	ASQ-3 Starter Kit: \$295.00 Includes: ASQ-3 User's Guide, ASQ-3 Quick-Start Guide, and a photocopiable print master set of 21 questionnaires and scoring sheets, as well as a CD-ROM with printable PDF questionnaires	The Denver II test kit is no longer available for purchase. The Training Manual and Test forms can be downloaded online.	Health Care Starter Pack (HC-S) \$150.00 Includes 75 IDI, 75 CDI-PQ, 75 CDC - With all necessary manuals and instructions.	PEDS-DM plus PEDS: \$346.00 Includes the PEDS-DM starter kit plus 100 PEDS Response Forms and the PEDS Brief Guide. The PEDS-DM Recording Form (100 supplied with each order) includes the PEDS Scoring/Interpretation Form that identifies when the PEDS-DM is needed.	The SWYC is available for free on the Floating Hospital for Children website.
<b>Ongoing costs</b>	None. All forms may be photocopied or printed from CD-ROM by the purchasing organization for no extra charge	None.	\$45.00 per pad of 75 (instructions included)	PEDS Response Forms (pad of 50): \$19.50 PEDS Score/Interpretation Forms (pad of 50): \$19.50 PEDS-DM Recording Forms (100): \$57.00	None.
<b>Languages</b>	English, Spanish, Arabic, Chinese, French, and Vietnamese	English and Spanish	English, Spanish, and Vietnamese	English and Spanish. Additional languages available to license.	English, Spanish, Khmer, Burmese, Nepali, Portuguese, Haitian-Creole, and Arabic
<b>Training options</b>	DVD training: ASQ-3 Scoring and Referral, Ages & Stages Questionnaires on a Home Visit; on site customized seminars offered through Brookes On Location professional development program;	Instructions for scoring and interpreting the PDQ II are found within the Training Manual	Instructions are included in the required Child Development Review Manual	Using PEDS: DM instructional video, PowerPoint slide shows, and additional downloadable training material available on website	Instructions are included in the SWYC User's Manual, available online.





# Mchat

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- <https://mchatscreen.com/>
- 20 question yes/no screening for autism
- Negative screening- score of 0-2
- Intermediate score of 3-7, administer the MCHAT follow up
- Sensitivity and specificity [ pooled data ] :
  - sensitivity of M-CHAT was 0.83 (95% CI, 0.77-0.88),
  - specificity of M-CHAT was 0.94 (95% CI, 0.89-0.97).
- Billing: CPT code 96110

## 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?<br>( <b>FOR EXAMPLE</b> , 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? ( <b>FOR EXAMPLE</b> , 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? ( <b>FOR EXAMPLE</b> , furniture, playground equipment, or stairs)  | Yes | No |
| 5. Does your child make unusual finger movements near his or her eyes?<br>( <b>FOR EXAMPLE</b> , 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?<br>( <b>FOR EXAMPLE</b> , 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?<br>( <b>FOR EXAMPLE</b> , pointing to an airplane in the sky or a big truck in the road)                  | Yes | No |



## Practical realities of screening

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- Can be done in academic centers or busy offices
- work with the staff to create a tailored workflow
- SMART goals- don't try everything at once
- ASQ and MCHAT can be done in waiting room- or prior to arrival
- Reimbursable

Specific, Measurable, Achievable, Realistic, and Timely.

## Doctor i'm concerned about my child....

---

- History
- Pregnancy issues:
- Birth history - premature ?
- Hearing and vision concerns?
- Any family history of slow growth?
- Is the delay sudden or gradual?
- Any regression ?
- Trauma or any change of social-family- school status?



## Autism DSM diagnosis

— — —

**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.
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.

## Autism diagnosis continued

— — —

**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).



## Autism diagnosis continued

— — —

C. Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities.)

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.

## If it's not autism

— — —

- **Anxiety disorder** – Anxiety disorder (includes social anxiety disorder, specific phobia, and selective mutism) has behavioral features that overlap with ASD.
- **Attachment disorder** – children with **severe early deprivation** or reactive attachment disorder may have abnormalities in social interaction, communication, and behavior.
  - Think trauma as well
- **Attention deficit hyperactivity disorder** – Children with ADHD may have impaired social function, though the impairments can be milder than those in children with ASD.
  - children with ADHD usually have normal pragmatic language skills, nonverbal social behavior, and imaginary play.





## If it's not autism

— — —

- **Global delay/intellectual disability**
  - Cognitive deficits can be difficult to assess in children
  - social responsiveness and communication of children with isolated ID is usually appropriate for their developmental level
- **Hearing impairment** –
  - *children with hearing impairment usually have normal reciprocal social interactions, imaginative play, normal eye-to-eye gaze, and facial expressions indicative of their intention to communicate*
  - **"trying but can't"**
- **Intellectual giftedness** –
  - "too smart for the class".
  - typically enjoy social interaction, have normal pragmatic language skills, **and can explain their intense interests**

## If it's not autism

— — —

- **Language disorder** –
  - children with developmental language disorder have normal reciprocal social interactions, normal desire and intent to communicate, and appropriate imaginative play
- **Language-based learning disorder** –
  - children with language-based learning disorders have normal reciprocal social interactions, normal desire and intent to communicate, and appropriate imaginative play.
  - Children with language-based learning disorder have difficulty/delay in **processing** content.
  - In addition, they want to communicate, but don't have the competency for it. **"trying but can't"**



## If it's not autism

---

- **Nonverbal learning disorder** –
  - Children with nonverbal learning disorder may have impaired social reasoning, strong rote skills, and well-developed language skills,
  - children with nonverbal learning disorder lack restricted, repetitive patterns of behavior, interests, or activities and usually have milder impairments in social skills and pragmatic language than those with ASD.
- **Obsessive-compulsive disorder** –
  - Children with OCD find their obsessions distressing
  - Conversely, children with ASD typically don't have insight
- **Rett syndrome** – Rett syndrome is a neurodevelopmental disorder that occurs almost exclusively in females.
  - Affected patients initially develop normally if head deceleration at about 3-6 months
  - gradually lose speech and stereotypical 'hand wringing'
  - Most cases of Rett syndrome result from mutations in the *MECP2* gene ( see genetics slide)

## If it's not autism

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- **Social (pragmatic) communication disorder** –
  - social (pragmatic) communication disorder is characterized by persistent difficulties in the social use of verbal and nonverbal communication
    - absence of restricted, repetitive patterns of behavior, interests, or activities.
- **Tic disorder/Tourette syndrome** – Similar to children with ASD, children with tic disorder or Tourette syndrome have sudden, brief, intermittent movements or utterances. Children with tic disorder or Tourette syndrome usually have normal social and communication/language skills.
  - Atypical social interactions in children usually related to the tics



## If it's not autism- less considerations

— — —

- **Fetal alcohol syndrome** –
  - a. Characteristic facial features of FAS (ie, short palpebral fissures, thin vermilion border, and smooth philtrum)
- **Landau-Kleffner syndrome** –
  - i. loss of previously established language milestones- normal development until 3-6 years old
  - a. inability to comprehend the spoken word
  - b. Can have seizures, but not required for diagnosis
- **Stereotypic movement disorder** – Similar to children with ASD, children with stereotypic movement disorder have repetitive, purposeless motor behaviors (eg, hand flapping, head banging) that may result in self-injury
  - a. Normal social interaction
  - b. Think - kid with parkinson's?

## Physical examination clues

— — —

- Weight – children with autism can have restricted diets
- Head circumference, including head circumference trajectory
  - 25% of ASD often have early acceleration of head growth,
- Approximately 15 percent of children with ASD have microcephaly
- Derm: hypopigmented macules of tuberous sclerosis complex
- Gen exam: dysmorphic features



## Labs

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- Hearing and vision
- Lead- particularly if living in old home
- Anemia
- No conclusive biomarkers
  - [Modern Biomarkers for Autism Spectrum Disorder: Future Directions | SpringerLink](#)

## Imaging ?

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Is a CT or MRI required?

Risks vs benefits to consider:

Sedation, Child might not comprehend what's going on,  
radiation exposure

Not routinely done, unless indicated by history and physical  
exam findings





## Genetics

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- Genetic studies of twins:
  - 98% concordance rate for monozygotic twins
  - 67% rate for dizygotic twins
- ASD recurrence rate
  - 10% for siblings
  - 25% if two siblings have autism

## Genetics of autism

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- No specific genetic mutation
- More than 100 genes linked to autism
- No variant accounts for more than 1% of cases
- Genetic evaluation is recommended AFTER DIAGNOSIS, not as part of the Diagnosis



## Genetics Methodology

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- Chromosomal Microarray analyzes a person's DNA for changes in the number of copies of certain segments of DNA, known as copy number variants (CNVs).
  - Some Deletions or duplications in chromosomes : 16p11.2, 15q11-q13, 22q11.2, 1q21.1, 7q11.23
  - CNVs can occur in healthy children and dont always indicate autism
  - Children with autism may not have any CNVs
- Whole genome sequencing is more comprehensive, and anylses both coding and non-coding regions.
- WGS can detect a wider range of genetic variants such as:
  - single nucleotide polymorphisms (SNPs)
  - small insertions and deletions (indels)
  - structural variants (SVs)
  - CNVs

## Based on this presentation, how do you diagnose Autism?

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- **Developmental history and direct observation.**
- All other testing is supportive.



## Partnering with Parents

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- Some parents will have concerns about the child's development, while the child's development is normal
- Conversely, what if a parent dismisses your concerns?
  - Stigma?
  - Cultural issues?
  - Explain that a referral to I.E or developmental doctor doesn't mean we are diagnosing with autism [same as with other specialist referrals]

## Autism Comorbidities- your role

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- Constipation
- Food selectivity
- Sleep disruption
- Educational placement and advocacy
  - Is a D75 school the best option for this child?
- Coordination with neuro/psych/development



## Take home points

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- Surveillance and screening CAN be done in the office setting!
- Hearing and vision are important to check.
- A good history will help you assess for medical issues or treatable diseases while you wait for a developmental doctor.
- Labs and imaging: focused testing based on H&P.
- Genetics are an adjunct- not diagnostic for autism.
- Our job doesn't end once a diagnosis is made.

## Resources

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- [ASQ- 9 month](#)
- [SQ 48 month sample](#)
- [M-CHAT](#)





## References

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- [Autism, Language Disorder, and Social \(Pragmatic\) Communication Disorder: DSM-V and Differential Diagnoses](#)
- [Clinical genetics evaluation in identifying the etiology of autism spectrum disorders: 2013 guideline revisions](#)
- [Evidence-Informed Milestones for Developmental Surveillance Tools](#)
- [Modern Biomarkers for Autism Spectrum Disorder: Future Directions | SpringerLink](#)
- [PEDS IN REVIEW Autism-Spectrum-Disorder](#)
- [Sensitivity and Specificity of the Modified Checklist for Autism in Toddlers \(Original and Revised\): A Systematic Review and Meta-analysis](#)



## Next steps: Diagnosis, Prognosis, Treatment and Family-Centered Care



May 5, 2023  
Harris Huberman MD

LEND



DOWNSTATE  
HEALTH SCIENCES UNIVERSITY

Disclosures:

**None**

LEND



DOWNSTATE  
HEALTH SCIENCES UNIVERSITY



1. Making the Diagnosis of ASD
2. Communicating the diagnosis of ASD and discussing prognosis
3. Treatments and next steps

## Making the Diagnosis of ASD

**Autism can be diagnosed from the age of \_\_\_\_\_.**

- a. 2 years
- b. 14 months
- c. 3 years
- d. 18 months



## Making the Diagnosis of ASD

Autism can be diagnosed from the age of \_\_\_\_\_.

- a. 2 years
- b. 14 months
- c. 3 years
- d. 18 months

## Making the Diagnosis of ASD

The diagnosis of Autism Spectrum Disorder (ASD) is based on \_\_\_\_\_.

- a) Genetic testing
- b) Taking a history
- c) Brain imaging
- d) Psychometric testing
- e) Direct observation
- f) a and b
- g) b and e





## Making the Diagnosis of ASD

The diagnosis of Autism Spectrum Disorder (ASD) is based on \_\_\_\_\_.

- a) Genetic testing
- b) Taking a history
- c) Brain imaging
- d) Psychometric testing
- e) Direct observation
- f) a and b
- g) b and e

## Taking a developmental history

- Autism Diagnostic Interview (ADI-R) – gold standard, 90-150 min, ages >2yrs
- Social Communication Questionnaire (SCQ) – brief (10'), ages >2yrs
- Social Responsiveness Scale (SRS-2) – 15-20', ages >2.5yrs
- Toddler Autism Symptom Inventory (TASI) – 40', ages 1-3yrs
- Childhood Autism Rating Scale (CARS-2) Parent Questionnaire

Hyman SL, Levy SE, Myers SM (2020) Identification, Evaluation and Management of Children with ASD. *Pediatrics* 145(1):e20193447  
Coulter et al (2021) The TASI: Use in diagnostic evaluations of toddlers - *Autism*



## Taking a developmental history

### ○ Develop your own history form -

5. "Before \_\_\_ could use words, say 14-16 months, how did he/she indicate his/her needs? \_\_\_\_\_  
Did he lead you to things? \_\_\_ Did he look back at you when he did? \_\_\_  
Did he ever put your hand on something as if your hand was supposed to make it happen?" \_\_\_ Example:  
Did he point to things he wanted? \_\_\_ When did he first point to desired things? \_\_\_
6. "What about pointing just to share his interest – say, to a bird or a bus to get you to look at it?  
Does he do this now? \_\_\_ When did he first do it?"
7. "What about gestures? Now, does \_\_\_ nod 'yes'? \_\_\_ When did he/she first nod? \_\_\_  
Now, does \_\_\_ shake head 'no'? \_\_\_ When did he first shake head no? \_\_\_  
Now, does \_\_\_ wave 'bye-bye'? \_\_\_ When did he first wave 'bye-bye'? \_\_\_  
Now, does \_\_\_ lift arms to be picked up? \_\_\_ When did he first lift arms? \_\_\_  
"Did \_\_\_ usually look you in the eye? \_\_\_ How is \_\_\_'s eye contact now? \_\_\_
8. "Does \_\_\_ imitate things you do?" (e.g. talking on the phone, sweeping?)  
When did he first imitate? \_\_\_ Please give examples: \_\_\_\_\_

Excerpt from Devel History form used at SUNY Downstate Child Devel Clinic

## Direct Observation

- **Screening Tool for Autism in Toddlers (STAT)** – 20', ages 2-3yrs
- **Communication & Symbolic Behavior Scales (CSBS-DP)** – Behavior Sample 30', 6mo-2yrs
- **Childhood Autism Rating Scale (CARS-2)** – history and observation
- **Autism Diagnostic Observation Schedule (ADOS-2)** – gold standard, 40-60', 12mo - adulthood

Stone, Coonrod, Ousley (2000) Brief report: screening tool for ASD in 2 year olds – *J Aut Dev Disord*  
Weatherby, Prizant (2002) CSBS-DP First Normed Ed - APA PsycNet



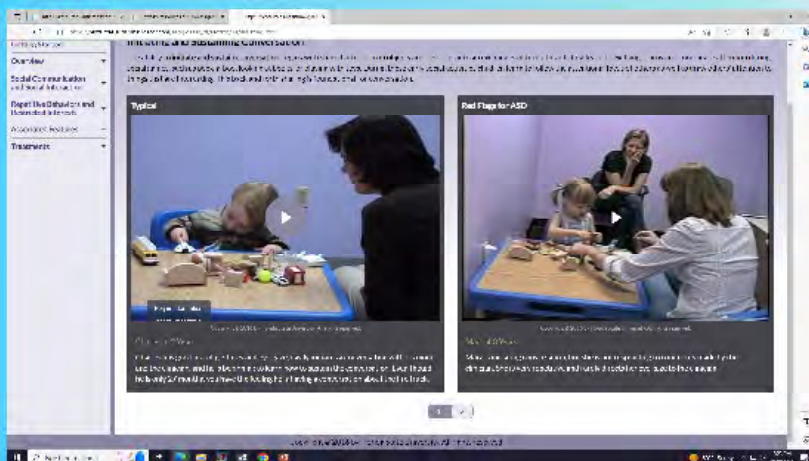
## Diagnosis of ASD

Selected clips from Autism Diagnostic Observation Schedule (ADOS-2) sessions

Autism Navigator – <https://autismnavigator.com/>

<https://resources.autismnavigator.com/asdglossary/#/section/11/initiatingConv>

#2 Charles 2 – Mara 3



## Making the diagnosis – concluding points

- CARS-2 – for more straightforward cases
- ADOS-2 – either when Dx is unclear (or need to “bring along a doubtful parent”)
- Importance of doing at least one formal diagnostic test (to support child receiving appropriate placements and ABA, OPWDD services)
- Regardless of formal score on CARS-2 or ADOS, need to step back, integrate all developmental history, current status (e.g. school performance, friendships) as well as CARS / ADOS test scores, and make a clinical judgment





**In your primary care clinic, you see a 30-month-old with speech delays, scores in the risk range on the MCHAT, and who is very self-directed and difficult to engage in your primary care clinic. The family has limited resources but is concerned and wants to know the best way to support their child. The current waitlist for an autism evaluation at the closest tertiary diagnostic center is nine months.**

**What is the BEST course of action?**

- a. Provide a preliminary diagnosis based on the MCHAT results and refer to early intervention services.
- b. Refer to the tertiary diagnostic center for autism evaluation and place an immediate referral for EI services.
- c. Repeat the MCHAT at 36-month follow-up visit.
- d. Encourage the family to monitor his development for the next six months using the CDC milestone tracker and come back in to reassess.
- e. Utilize the DSM-5 criteria, score a CARS-2 based on your history and observations, and make the diagnosis of ASD yourself

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## Family Centered-Care and communicating the diagnosis

What is “family centered-care”?

- a. Decision-making based on a partnership between provider and family
- b. Appreciation and respect for diversity, cultural and linguistic traditions and care preferences
- c. Care in context of family, school, community

How does family centered-care play out in approaching the diagnosis of ASD or other neurodisability with the family?



## Communicating the diagnosis

It is imperative to consider all of the following when discussing ASD diagnosis and next step clinical recommendations with caregivers during a feedback session EXCEPT \_\_\_\_\_.

- a. Potential risk factors of autism that caregivers may have contributed to.
- b. Impact of race, ethnicity and cultural background on understanding of autism.
- c. Factors related to the family's socioeconomic status and their influences on access to recommended services.
- d. Caregiver's learning abilities and skills related to service navigation and advocacy.

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## Communicating the diagnosis

- Point out the positives about the child
- Explain the meaning of the ASD diagnosis and “spectrum”
- Discuss how the diagnosis should ‘open doors’ and what next?

## Communicating the diagnosis

### The meaning of the ASD diagnosis and “spectrum”

- ASD is ‘not one thing’, extremely heterogeneous (and individuals vary tremendously in terms of intelligence, engageability, focus, anxiety, rigidity ... and of course with their own personality)
- but with shared features
- ... that will hopefully help the family and others understand the child / young person





## Communicating the diagnosis

Anticipate questions such as:

- What causes ASD?
- Is my child “high- or low-functioning”?
- What will be her/his future?

## Communicating the diagnosis

What causes ASD?

- That its ‘neurobiologically-based’ or ‘differences in how the brain is wired’, (and *not* how you raised them), is highly heritable (but not in simple genetic fashion...)
- ... but explain that there are likely many different causes that contribute to ASD
- (but vaccines aren’t one of them!)
- and acknowledge that neither the medical nor the research community has a clear answer





## Communicating the diagnosis

Is my child “high- or low-functioning”?

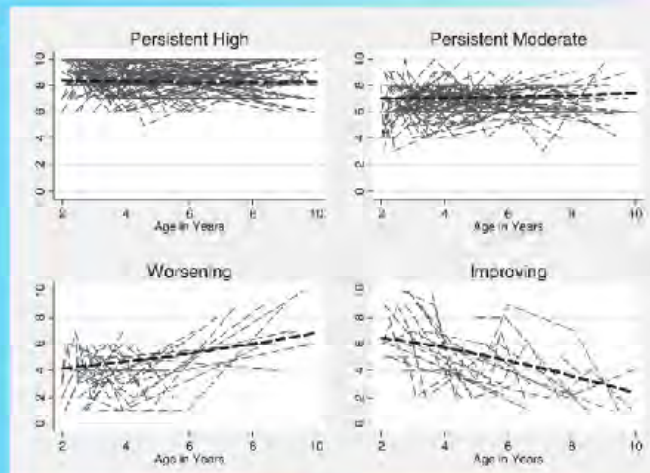
- Terminology – moving away from ‘high / low functioning’
- Refer to the DSM-5 severity levels:
  1. Requiring support (in social communication / RRB’s)
  2. Requiring substantial support ( “ “ / “ )
  3. Requiring very substantial support ( “ / “ )
- These refer to current functioning, not the future

## Communicating the diagnosis

What will be her/his future?

- At an individual level ... we don’t know

- At a population level –  
Cathy Lord study with  
serial ADOS for 10 yrs



- Generally, we expect most children to make substantial progress in language, social interaction and learning



## Terminology – moving away from ‘high / low functioning’

### A vignette:

*Felipe was initially seen in Devel Clinic at age 4 and diagnosed with ASD based on his limited eye contact, “liking to have people around, but he’s not with [people]”, fascination with doors and subways, and self-directed play. He made good progress in his language and social skills in a special ed preschool and later in a ‘learning differences’ school, beginning to interact verbally and make friends.*

*Now 15 year old, Felipe has become a charming teenager with a somewhat idiosyncratic way of speaking and an impish sense of humor. He participated in a LEND Workshop of Self-Advocates speaking about being on the spectrum.*

*He spoke against the notion of a cure for autism, and “if I had the choice of not being on the spectrum, I would say no, I’d still be on the spectrum, because it made me who I am”*

## Terminology – moving away from ‘high / low functioning’

Felipe’s comments reflect the thinking of many in the ASD community who see autism as a form of ‘neurodiversity’ and to be seen as a positive part of the human experience ... and argue (especially with the medical community) that we should stop using words that connote impairment or pathology, like ‘disorder’, ‘problem behaviors’, ‘treatment’ or ‘prevention’.

We may question whether this viewpoint speaks for all people on the spectrum, but it’s a valuable counterpoint to our medical perspective, and one you should try to convey in presenting an ASD diagnosis to a family.

The problem with ‘high functioning’ or ‘low functioning’ terms is that they fail to capture the different dimensions and complexity of individuals with ASD





## ASD – Treatments and Next Steps

- Main treatment is combination of educational programs and therapeutic treatments

Hyman SL, Levy SE, Myers SM (2020) Identification, Evaluation and Management of Children with ASD. *Pediatrics* 145(1):e20193447

- Therapeutic treatments include: Speech Therapy, Occupational Therapy, Physical Therapy,
- Strongest, evidence-based Applied Behavioral Analysis (ABA)
- 0-3 years through Early Intervention
- Specialized school programs (e.g. TEACCH), in NYC range from ASD Nest inclusion classes to small (6:1:1), self-contained classes in District 75 schools
- What about medical) treatments for ASD?

## Treatments and Next Steps

**Medications approved to treat the *core* features of ASD include \_\_\_\_\_.**

- a. Guanfacine
- b. Aripiprazole
- c. Methylphenidate
- d. Risperidone
- e. Sertraline
- f. All of the above
- g. None of the above



## Treatments and Next Steps

Medications approved to treat the *core* features of ASD include \_\_\_\_\_.

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- d. Risperidone
- e. Sertraline
- f. All of the above
- g. **None of the above**

## Treatments and Next Steps

Target symptom	Medication Class
Hyperactivity, impulsivity, inattention, distractibility	Stimulants (methylphenidates, amphetamines) SNRI's (atomoxetine) $\alpha$ -2 agonists (guanfacine, clonidine)
Irritability, severe disruptive behavior	$\alpha$ -2 agonists (guanfacine, clonidine) Atypical antipsychotics (aripiprazole, risperidone) SSRI's (fluvoxamine, citalopram) Mood stabilizers (e.g. valproic acid)
Repetitive, rigid behaviors	Atypical antipsychotics (aripiprazole, risperidone) Mood stabilizers SSRI's
Anxiety, depression	SSRI's $\alpha$ -2 agonists (guanfacine, clonidine) Atypical antipsychotics (aripiprazole, risperidone)

Table summarized from:

Hyman SL, Levy SE, Myers SM (2020) Identification, Evaluation and Management of Children with ASD. *Pediatrics* 145(1):e20193447



## ASD – Treatments and Next Steps

Guiding family through:

- Accessing ABA, other therapies through insurance
- Working with the school system, changing educational classification on IEP, advocating for the right placement, ancillary services
- Accessing services through NYS Office of Persons with Developmental Disabilities (OPWDD)
- Obtaining Medicaid Waiver
- As a provider, you yourself won't do these things – but you need to know who to call – case managers, support programs
- One of the places where the Social Worker becomes the key member of the team!

## ASD – Treatments and Next Steps

Importance of the larger systems of care and networks of programs

- Helping families negotiate their way through
- LEND – interdisciplinary training to help providers
- LEND - Family Support Project:
  1. Emotional support
  2. Guidance on systems of care
  3. Advocacy Training
- We'll hear more from upcoming Panels, the afternoon Roundtable and program materials with additional information and resources





Thank you

LEND



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