Autism Across the Lifespan

Rose Mary Merola M.D.
Neurodevelopmental Pediatrician
Disclosures

- Neither I nor any of my immediate family has a financial relationship or interest with any proprietary entity producing health care goods or services related to the content of this activity.

- My content will not include discussion or reference of any commercial products or services.

- I do not intend to discuss an unapproved or investigative use of commercial products or devices.
Objectives

- Discuss the core symptoms; associated clinical features, developmental and behavioral difficulties that need to be addressed
- Discuss treatment options for children over the lifespan
- Discuss outcome
Neurodevelopmental Pediatrician

- Pediatrician
  - Post completion of 3 year pediatric residency program
- Fellowship in Development and Behavioral or Neurodevelopmental Pediatrics
  - Post completion of 2 year Fellowship Program
  - Board Certified in Neurodevelopmental Disabilities
- Evaluation of children from Birth to 21yo with all types of Developmental and/or Behavioral Problems
DSM - V : Current Concept of Autism

- Behavioral disorder or syndrome
  - Core symptoms present in all children on the spectrum
  - Each child may present with different degrees of the core symptoms even within the same family
  - Sensory Integration Dysfunction added to DSM V as part of presentation
  - ADL/life skills independence is used to determine severity of the condition

- Multiple etiologies: Genetic/Environmental

- Lifelong disorder
  - Different appearance (e.g., peer interactions change through life)
  - Importance of early diagnosis for best outcome
  - Need for sustained support

- Selective or greater impairment in social interaction
**DSM-V Criteria for Autism Spectrum Disorder**

- Currently or by history must meet the criteria A, B, C and D
- A. Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, and manifest by all 3 of following:
  - Deficits in social-emotional reciprocity
  - Deficits in nonverbal communicative behaviors used for social interaction
  - Deficits in development and maintaining relationships
- B. Restricted, repetitive patterns of behavior, interests or activities as manifested by at least 2 of the following:
  - Stereotyped or repetitive speech, motor movements or use of objects
  - Excessive adherence to routines, rituals of verbal/nonverbal behavior or need for sameness
  - Highly restricted, fixated interests that are abnormal in intensity or focus
  - Hyper or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment
- C. Symptoms must be present in early childhood (but may not fully manifest until social demands exceed limited capacities)
- D. Symptoms together limit and impair everyday functioning
SOCIAL DEFICITS

► **JOINT ATTENTION (Amygdala)**
  - Universal and specific to autism
  - Inability to coordinate one’s own attention between and object and another person to:
    - Indicate a need (Proto - Imperative Pointing)
    - Share an interest (Proto - Declarative Pointing)
  - **DEVELOPMENT OF JOINT ATTENTION USUALLY PREDICTS ONSET OF FUNCTIONAL LANGUAGE WITHIN A YEAR**
  - Amygdala is the emotional center
Joint Attention

Definition:
- It is the ability to share a common focus on something with someone else
  - Common focus on: PEOPLE / OBJECTS / CONCEPT / EVENT ...

Involves: ability to gain, maintain and shift attention

Serves: it is a reference tool, that uses shared gaze (visually focusing on same object), and/or gesture for communication.
- Sharing a focus
  - not only helps develop communication skills
  - Also helps develop important social skills
    - Bonding
    - Seeing another’s point of view

Predictor: it is a predictor of future language development
- Once joint attention is established, language will follow within a year!
Joint Attention

Early signs

- 2mos  reciprocal smiling
- 5mos  attachment to caregiver
- 8mos  gaze monitoring
- 10mo  follows a point
- 12mo  Proto-Imperative pointing
  - Pointing and looking at parent to obtain an object
  - Reaching to be picked up
- 14mo  Proto-Declarative pointing
  - Vocalizing and pointing, prompting parent to look at object obtained
  - Shows object with eye contact
  - Looking at same page in a book

Later signs

- Focusing on a game
  - > 3yo playing a board or card game
- Playing make believe
  - > 2yo - Common place pretend
  - > 3yo - Associative play
  - > 4yo - Role play
- Requesting items desired
  - Food /Drink
  - Toy
  - Going outside / Going home
Skills Needed for Joint Attention

- Orienting and attending to a social partner
- Shifting gaze between people and objects
- Sharing emotional states with another person
- Following the gaze and point of another person
- Being able to draw another person’s attention to objects or events for the purpose of sharing

- THESE SKILLS NOT ONLY ASSIST THE CHILD TO SECURE WANTS/NEEDS
- BUT, THEY ARE NEEDED FOR DEVELOPMENT OF APPROPRIATE SOCIAL INTERACTIONS AND FOR DEVELOPMENT OF MEANINGFUL RELATIONSHIPS
How to Improve Joint Attention

- Parent needs to be a good language role model
- Be sure to have consistent routines, especially with daily living skills
- Practicing joint attention in the natural environment, helps the child to achieve communication and social success at home, at school and in the community
- Use gestures: POINTING WITH EYE GAZE to show the child where to direct his/her focus
- Use gestures: HAND OVER HAND to direct the child to mimic the gesture
  - Actually take the child’s hand and help him/her point to an object to practice gestures
  - Begin with an object the child is interested in to start
  - When the child shows interest in something - You can mimic the interest
  - Add a comment when pointing
    - “You want the Truck? The Big Red Truck?
  - Add a gesture with a visual cue
    - Point to the Truck, then Point to your eye and Draw a Pretend Line going from your eye to the object/truck
SOCIAL DEFICITS

- THEORY OF THE MIND (Frontal Cortex, Amygdala)
  - Lack of realization that others have thoughts or emotions independent from one’s own emotions
  - “Mind Blindness”
  - Inability to take the perspective of another
  - Inability to pass False Belief Tasks
Frontal lobe
Executive functions, thinking, planning, organising and problem solving, emotions and behavioural control, personality

Motor cortex
Movement

Sensory cortex
Sensations

Parietal lobe
Perception, making sense of the world, arithmetic, spelling

Occipital lobe
Vision

Temporal lobe
Memory, understanding, language
Sensory Integration Dysfunction

- Occurs when the brain is not processing or organizing the flow of sensory impulses in a manner that gives the individual good, precise information about himself or his world.

- When the brain is not processing sensory input well, it usually is also not directing behavior effectively.

- Without good sensory integration, learning is difficult and the individual often feels uncomfortable about himself, and cannot easily cope with ordinary demands and stress.
Sensory Integration Disorder

- **Tactile**
  - **Feeding**
    - Gags on textures
    - Poor chewing
    - Poor suck/swallow
    - Don’t mix foods
    - Color specific
  - **Clothing**
    - Refuses certain textures
    - Difficulty changing clothes to match season
    - Refuses clothes altogether
  - **Hygiene**
    - Won’t wash or dry skin
    - Won’t wash hair
    - Doesn’t like to be wet

- **Auditory**
  - Hyper - Sensitivity to noises
  - Not sensitive to noise/seems deaf
  - Sensitive to criticism/scolding

- **Olefactory**
  - Sensitivity to smells/odors
  - Car sickness
Sensory Integration Dysfunction

**Vestibular - balance and coordination**
- Doesn’t get dizzy spinning
- Poor coordination and awkward at sports
- Feels heavier than same weight peer without sensory deficits
- Can’t hold up head/arms/legs at same time when on his stomach
- Hands and feet do not work well together
- May be ambidextrous/confused dominance
- May get right/left confused
- Does not tolerate stress well when frustrated
- May have reading and math difficulties and may reverse letters and words
Vestibular skills
Proprioceptive skills/deficits

Proprioception = Kinesthesia

We get sensory information from our muscles and joints telling our brain where that part of our body is located and how it is moving so we can plan the next action.

Imagine drinking a cup of coffee blindfolded

- How do you know:
  - Where your mouth is?
  - Where your hand is?
  - How heavy the cup is?
  - How fast you are moving?

You know these things through the sensations sent by your muscles and joints to your brain or proprioception.

If we did not have proprioception, we would have a terrible time completing even simple actions.
Examples of Proprioception
Sensory Integration Dysfunction
Symptoms

- Hyperactivity and Easily Distracted Child
  - Hyperactivity
    - Child is in constant motion
    - Child runs not walks
    - Most of his/her activity is non purposeful
    - It’s almost impossible for child to sit and concentrate
  - Distracted easily
    - Child may be unable to shutout noises/lights and confusion of many people doing different things which may cause fear of crowds, melt downs outside his routine
    - Young child is busy all the time or gets hyper-focused to shut out any distractions
    - Older child may slow down but cannot keep his/her room in order or get homework done or get to school with the things he/she needs
  - If a brain cannot organize sensory input and motor activity THEN it cannot organize a closet full of clothes or a satchel of books, paper or pencils
Sensory Integration Dysfunction
Symptoms

- **Behavioral Problems**
  - **Infant**: may be less happy, more irritable; wakens easily with noise; unable to be calmed; need to be held all the time
  - **Toddler**: may be fussy and not want to be around people, including family and peers; want to stay at home where he/she has regular routine; cries when others visit; wants to separate to play by him/her self; aggressive if bothered by peers
  - **Child**: may get very upset if things don’t go his/her way; can become aggressive if mom says ‘no’; if ipad stops working, may throw it; won’t share food or toys; overly sensitive to reprimand; feelings hurt easily; cannot cope with daily stressors; doesn’t like to try anything new or to be in new situations
  - May be overly emotional or have emotional dysregulation

- **Consequences of his/her behaviors**
  - Overly emotional children may *alienate themselves* from peers or *be rejected* by peers
  - Overly emotional child may *cause parent to lose control and react negatively* to his/her behavior
  - **Child is set up for negative self concept cycle and misery**
  - Child may prefer to play with older children/adults who understand his/her dilemma and be willing to work with him/her
Sensory Integration Dysfunction
Symptoms

Speech and Learning problems

Speech problems: speech depends on many sensory integrative processes
  - Total lack of expressive speech
  - Poor receptive language skills or poor processing of information
  - Repetitive but non functional speech development

Learning problems: if SI problem is mild you may not see problem until child enters school.

  - May see problem Reading / Writing / Math
    - Direct problem due to the brain or SI problem or
    - Indirect problem due to his lack of attention/behavior
  - Reading and Writing require memory of direction which his visual and muscular memory that is altered by sensory system dysfunction
  - Some children may have difficulty translating what they hear to the physical act of writing or taking aural sensation and moving it from hearing to motor tracts for writing
  - The child may have difficulty copying from the board, trouble with spacing of letters on paper and the direction the letters need to go
Sensory Integration Dysfunction
Symptoms

- **Muscle tone and Coordination**
  - Sensations from vestibular and proprioceptive systems provide muscle tone that keeps the body upright and energetic
  - Child with SE dysfunction has low muscle tone which makes him seem weak, and the effort needed to move against gravity, causes him to have low endurance and to give up more easily.
  - When vestibular, proprioceptive and tactile systems are not working well, poor motor coordination is seen
    - Child may lose balance and stumble easily
    - Child may drop his pencil more often than other children
    - Some children fall out of their chairs because they cannot feel exactly where they are on their seat
    - Clumsiness and immature play are common early signs
    - Can see difficulty: building blocks, manipulating toys and playing with puzzles
    - SI dysfunction may cause child to bump into things and people since he cannot judge where things are in space or where his body is in space.
Sensory Integration Dysfunction
Symptoms

- **Teen age problems**
  - Child may adapt as he/she grows or lose interest in school
  - If SI problems are mild/less severe
    - May have difficulty turning to correct side (L/R confusion)
    - May have difficulty remembering sequences of numbers
    - May have difficulty counting change
  - Adolescent with SI disorder may avoid dancing or have difficulty clapping out a rhythm to music
  - Adolescent may have lack of organization and focus on tasks
    - Unable to keep his/her room neat or clean room
    - Can’t organize to write a book report
    - Can’t decide what to do 1st
    - If interrupted forgets what he/she was doing
    - Unable to finish what he starts
Sensory Integration Dysfunction comorbidities

- **ADHD/attention deficit hyperactivity disorder**
  - Child does not have core symptoms of ASD/autism
  - May have sensory integration disorder/dysfunction
  - May have social interaction problems due to their impulsivity and SI disorder but good conversation skills

- **Learning disability, especially NVLD/non verbal LD**
  - “Right brain child”
  - Child has sensory integration dysfunctions
  - Social interaction issues/poor peer relationships due to hyperactivity and SI issues but good conversation skills
  - Math problems

- **Bipolar disorder (manic depressive disorder)**
  - Cyclical manic and depressive episodes
  - Flight of ideas
  - Narcissistic

- **Autism Spectrum Disorder**
  - Most intense
Communication Impairment of ASD

- Absent or delayed language without the attempts to compensate
  - 50% are non verbal
  - 25-30% regress between 1-2 yo
  - Inconsistent use of words, Neologisms
- When adequate language is present, see marked impairment in the ability to sustain a conversation
  - Limited functional language
- Stereotypic or repetitive use of language
  - Scripted or repetitive speech
- Lack of make believe, social imitative play
- Dissociation between:
  - Form = syntax/articulation (increased)
  - Function = pragmatics, social use (decreased)
Stereotypic Behaviors

- Restricted interests that are abnormal in focus or intensity
  - Hard comfort items (pen/rock/feather..) = Red Flag!
  - Fixations: puzzles/ leggos/ U tube videos / numbers / letters
  - Need for sameness
- Inflexible non-functional routines
- Pre-occupation with parts of objects
- Stereotypic motor mannerisms
  - Often not apparent until >3yo when social/communication demands exceed capabilities
MYTHS of ASD diagnosis

- No eye contact
- No speech
- Rare
  - 1:64 in New Jersey
- Behavioral etiology (Refrigerator Mother)
  - Not an attachment disorder
- Post-natal onset
  - felt to be due to genetics/environment during pregnancy
- No definitive diagnosis until child 4 yo
  - Diagnosis can be made by 18-24 months of age
- Cognitive Impairment > 90%
  - Actually only in 40-70 %
Science of 21st Century

- Not Rare
  - Occurs in 3-5 per 1000
- Prenatal Onset
  - Suspected at around 21 days gestation
- Multi-factorial Etiology
  - Strongly Genetic (95%)
  - Neuro-pathologic correlates
- Wide Spectrum of Phenotypes for ASD
  - 7-20 genes involved in presentation of behavioral phenotype
- Increased use of screening and diagnostic tools
- Early Intervention = Effective in Many children
Multifactorial Etiology of ASD

- Environmental (5-10%)
  - Toxins
    - Lead poisoning
    - Alcohol (in utero)
    - ?? Drugs especially marijuana/alcohol/cigarettes??
    - ??? Hormonal shifts during pregnancy??

- Medications
  - Retinoid
  - Thalidomide
  - Valproate

- Viruses
  - CMV
  - Rubella
Neurodevelopmental Assessment

- Complete history from birth to present age for risk factors
- Physical exam for medical problems or syndrome features
- Neurological examination for general or focal findings
- Developmental exam <5yo
  - 5 domains: motor/language/adaptive/cognitive/social
- Neuropsychological screen
  - Learning deficits/language deficits
  - Behavior evaluation using standardized scales

- History and Physical exam tells me how to proceed!
Developmental Evaluation

- Look for deviant developmental patterns
- Patchy skills especially language
  - Great rote memory skills but no conversational abilities
- Significant deficits in social and language areas compared to other domains
- Receptive language deficits may exceed the expressive deficits.

- Looking for strengths and weaknesses
Neurodevelopmental Exam - ASD

- Look for Associated Clinical Features:
  - Intellectual deficiency 40-70% dependent on severity ASD
  - LD/learning disability (CAPD/receptive or processing deficits/SI disorder)
  - Medical:
    - genetic (syndromes)
    - neurologic: seizures 20-25% with peak <3yo and > 13/puberty

- THESE MAY HAVE GREATER IMPACT ON SUCCESS AND OUTCOME THAN CORE SYMPTOMS
Medical Work-up for ASD

- Audiology baseline since most present with language deficits
- Genetic testing at time of first diagnosis all with ASD
- Any other tests depend on child’s presentation
Management of ASD

- EIP/early intervention services = Birth to 3 years of age
  - DI/developmental intervention
    - Play skills
    - Cognitive skills
    - Language skills until child able to say words

- Speech therapy
  - If too much or too little speech
  - Teach scripted speech, starting with single words/phrases/sentences
    - PECS - Picture Exchange Cards
    - Articulation deficits addressed as well
  - Social stories used to change negative behaviors and promote social skills learning
  - Speech groups for social communication training

- Occupational therapy
  - Sensory gym
    - swinging
    - Jumping
    - Bouncing
    - Ball activities
    - Spinning
  - Fine motor skills training
    - Fine motor dexterity/grasp
    - Drawing skills/ prewriting skills
    - Coordinated movements
  - Gross motor skills training
    - Scooter/ big wheels/ tricycle
  - OT/ABA for promoting attention and reduction of hyperactivity
<table>
<thead>
<tr>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
<th>PECS</th>
</tr>
</thead>
<tbody>
<tr>
<td>circle</td>
<td>triangle</td>
<td>square</td>
<td>rectangle</td>
<td>change clothes</td>
<td>fork</td>
<td>spoon</td>
<td>plate</td>
<td>cup</td>
<td>bowl</td>
</tr>
<tr>
<td>pencil</td>
<td>pencil bag</td>
<td>bag</td>
<td>yes</td>
<td>no</td>
<td>itchy</td>
<td>cough</td>
<td>shirt</td>
<td>shorts</td>
<td>trousers</td>
</tr>
<tr>
<td>sit down</td>
<td>quiet</td>
<td>wipe mouth</td>
<td>sneeze</td>
<td>yes</td>
<td>no</td>
<td>itchy</td>
<td>cough</td>
<td>shirt</td>
<td>shorts</td>
</tr>
<tr>
<td>belt</td>
<td>medicine</td>
<td>koosh ball</td>
<td>play</td>
<td>computer</td>
<td>penguin</td>
<td>bears</td>
<td>dog</td>
<td>sunny</td>
<td>raining</td>
</tr>
<tr>
<td>windy</td>
<td>cold</td>
<td>hot</td>
<td>noodles</td>
<td>cake</td>
<td>biscuit</td>
<td>chips</td>
<td>drink</td>
<td>juice</td>
<td>rice</td>
</tr>
<tr>
<td>chocolate</td>
<td>apple</td>
<td>fish burger</td>
<td>eyes</td>
<td>ears</td>
<td>I see</td>
<td>I hear</td>
<td>pink</td>
<td>grey</td>
<td>white</td>
</tr>
<tr>
<td>black</td>
<td>purple</td>
<td>blue</td>
<td>red</td>
<td>yellow</td>
<td>green</td>
<td>orange</td>
<td>ice cream</td>
<td>eat</td>
<td>sky train</td>
</tr>
<tr>
<td>wash</td>
<td>swim</td>
<td>aeroplane</td>
<td>shopping</td>
<td>church</td>
<td>shopping center</td>
<td>home</td>
<td>school</td>
<td>toilet</td>
<td>airport</td>
</tr>
<tr>
<td>playground</td>
<td>car</td>
<td>bus</td>
<td>train</td>
<td>bicycle</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Augmentative Communication Devices
OT gym
Behavioral Therapy

- **Applied Behavioral Analysis (certified ABA therapist)**
  - ABA is an applied science devoted to developing procedures which will produce observable changes in behavior.
  - **Functional Behavioral Analysis** is completed to determine behaviors to be reduced by determining their cause and frequency.
- **Contact Perform Care/DDD: 1-877-652-7624**
- **Contact Insurance with letter of medical necessity and prescription for ABA**

- **Behavior modification Therapy: (BCBA)**
  - Uses: A B Cs of behavior management
    - Antecedant / Behavior / Consequence
    - If child is avoiding activity by crying or melting down, ask the child to perform the activity by using a motivator as reward
  - Conditioning:
    - Use of rewards to increase the activity desired and punishments(extinctions) to reduce negative behaviors
  - Reinforcer
    - Food is the most common reinforcer
    - Time on Computer/Ipad or other devices can be used
    - TV time
IDEA: Management of ASD

- **Diagnosis 18-24mo (birth to 3yo)**
  - EIP: ABA/DI/ST/OT/PT
  - In home ABA to promote language and reduce negative behaviors

- **Diagnosis 3-5yo**
  - IFSP: ABA or behavior therapy/ST/OT/PT/Social skills
  - Social skills/ST groups
  - Social skills/ Lunch Bunch

- **Diagnosis > 5yo**
  - IEP: Self contained class vs Inclusion class vs general education with /without resource room pull out
  - PT/OT/ST/social skills/ behavior therapy
  - Special education school specific for autism/ASD
  - ABA if still indicated
**IDEA: Management of ASD**

Middle school/High school
- IEP depends on language abilities/IQ/Behavioral issues
- High school level: vocational training versus continued academic prep considered
- Social skills continue: school and privately

College
- Accommodations are needs based
- IQ may determine if child qualifies for college and which program is best

**WEBSITE: thinkcollege.net**
Autism Websites

- www.autismspeaks.org
- www.autism-society.org
- www.disablityscoop.com
- www.autismnj.org
- www.autism.com
- www.autismweb.com
- www.ahany.org
- www.autismhwy.com
- www.autismbeacon.com
- www.autism.healingthresholds.com
- Thinkcollege.net
  - Colleges that accommodate for special needs adults
Use of apps for development of social skills in autism

- See List of apps for social skills and ASD

- **Mission Rescue Kloog**
  - Tablets only
  - Teaches: children / adolescents / teens
  - 17 lessons each covering a required social skills
Music Therapy or Lessons

6 benefits of music lesson

- It improved academic skills, especially music and math
- It develops physical skills, especially percussion instruments
- It cultivates social skills
  - Group classes require peer interaction and communication which encourages teamwork
- It refines discipline and patience
  - Learning an instrument teaches children about delayed gratification
- It boosts self esteem
  - Lessons offer a forum where child can learn to accept and give constructive criticism
- It introduces children to other cultures
Key to Success

- **LOOK FOR STRENGTHS AND WEAKNESSES**

- **Early Diagnosis and Intervention**
  - Earliest diagnosis around 18 months of age
  - EIP with therapies specific for each child
  - ABA: in home and at daycare if indicated
  - Apps appropriate for learning and used with developmental therapy

- **Appropriate use of Behavioral Therapy with Rehabilitation Therapy**
  - Behavioral: ABA and Behavior modification
  - Rehabilitation: physical/occupational/speech therapies
  - Social skills: formal social skills groups

- **Adding complimentary therapies**
  - Music / Dance / Swimming / Gymnastics / Sports
  - Daily exercise for reducing stress levels
  - Play dates and group activities with peers and family