Autism Spectrum Disorders

— A Primer for Practitioners —

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Disclosures

I have no financial relationships or affiliations to disclose.
Learning Objectives

- Appreciate the clinical context and community impact of Autism Spectrum Disorder (ASD).
- Learn DSM-5 symptom criteria for ASD.
- Identify the early signs of ASD.
- Understand screening and assessment.
- Learn about the various evidenced-based interventions.
Introduction

- Autism was first described in 1943 by Leo Kanner.
- He reported on 11 children ...
  - Congenital inability to relate to other people
  - Sensitive to changes in the environment
  - Lack of interest in people
  - If language developed, it was marked by echolalia, pronoun reversal, concreteness
  - Exhibited unusual, repetitive, and apparently purposeless activities (stereotypies)
Clinical Context: Prevalence

- ASD is the 5th most prevalent pediatric mental health disorder in the United States.

- Rising prevalence rates
  - 1943: 2-4 per 10,000
  - 2006: ~1/110
  - 2010: ~1/68

- About 4 times more common in boys than girls.

- Females with ASD tend to have more severe intellectual disability.
Clinical Context: Impact

- The lifetime cost of a person with ASD and an intellectual disability averages $2.4 million, and $1.4 million without an ID.

- The yearly cost to the United States is an estimated $236 billion a year.

- Autism severity is a main driver of costs across the lifespan ...

- ... the key to reducing costs includes better access to early detection & quality intervention.
Clinical Context: Correlates

- 10% with identifiable genetic and chromosomal etiologies (eg, Fragile X Syndrome, tuberous sclerosis, metabolic syndromes).
- 90% *idiopathic* with multiple genetic determinants and environmental factors.
- 20% have electroencephalographic (EEG) abnormalities and seizure disorders.
- ~40% diagnosed with ASD have an Intellectual Disability (ID).
Clinical Context: Some Neurobiology

- Postmortem studies have shown various abnormalities, particularly within the limbic system.
- fMRI procedures have identified difficulties in tasks involving social & affective judgments, and differences in the processing of facial and non-facial stimuli.
- Frequently replicated neurochemical finding is elevation of peripheral levels of the neurotransmitter serotonin.
Clinical Context: Neuropsychology

- Impairments in executive functioning (simultaneously engaging in multiple tasks).
- Weak central coherence (integrating information into meaningful wholes).
- Deficits in theory-of-mind tasks (taking the perspective of another person).
Clinical Context: Familial Factors

- Higher rates of autism are consistently noted in siblings of affected children.
- Identified risk factors for ASD appear to include:
  - closer spacing of pregnancies
  - advanced maternal, or paternal age
  - extremely premature birth (<26 weeks GA)
- High rates of learning/language problems and social disability in family members.
- A possible increase in the risk for mood/anxiety disorders has been noted in family members.
Clinical Context: Early Considerations

- Some ASD children show normal development until around 18 to 24 months of age; then stop gaining new skills, or lose the skills that were developed.

- Studies show 1/3 to 1/2 of parents with an ASD child had concerns before the 1st birthday.

- Nearly 80%–90% of parents had concerns about their child by 24 months.
Clinical Context: Early Considerations

- ASD can be reliably diagnosed in children less than 3 years old.
- Multiple studies suggest the diagnosis of autism when made in younger children remains stable over time.
  - Ozonoff et al (2014)
- Yet, most children are not diagnosed until at least 4 years old.
Clinical Context: Access

- There is huge shortage of specialty providers in pediatric mental health across the U.S.
- Less than 7,500 practicing Child and Adolescent psychiatrists.
- Only about 600 Developmental Behavioral Pediatricians in the U.S.
- Shortages of providers often means significant delays in access.
DSM 5 Diagnostic Criteria

- **Criterion A.** Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history
  - A1. Deficits in social-emotional reciprocity
  - A2. Deficits in nonverbal communicative behaviors used for social interaction
  - A3. Deficits in developing, maintaining, and understanding relationships
DSM 5 Diagnostic Criteria

- Criterion B. Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least 2 of the following (currently/by hx)
  - B1. Stereotyped, or repetitive motor movements, use of objects, or
  - B2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal
  - B3. Highly restricted, fixated interests that are abnormal in intensity or focus
  - B4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects of the environment
DSM 5 Diagnostic Criteria

- **Criterion C.** Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities).

- **Criterion D.** Symptoms together limit and impair everyday functioning.

- **Criterion E.** These disturbances are not better explained by intellectual disability or global developmental delay.
The Many Complexities of ASD
What are the Early Signs of ASD?
Early Signs: The “Red Flags”

**Typical Developing Toddlers versus ASD Toddlers**

**Demonstration Video**

- ASD typically appears during the early years of life.
- Early assessment and intervention are crucial to a child's future success.
- Talk to your pediatrician about concerns.

**Early Signs**

- **No social smiling by 6 months**
- **No one-word communications by 16 months**
- **No two-word phrases by 24 months**
- **No babbling, pointing, or meaningful gestures by 12 months**
- **Poor eye contact**
- **Not showing items or sharing interests**
- **Not responding to sounds, voices, or name**
- **Loss of skills at any time**
- **Unusual attachment to one particular toy or object**
Bringing the Early Signs of Autism Spectrum Disorders into Focus
All children should receive periodic developmental screening using a standardized test.

In the absence of established risk factors or parental or provider concerns, a general developmental screen is recommended at the 9, 18, and 24 month visit.

If screening results are concerning, the child should be scheduled for developmental or medical evaluations.
## Select Screening Tools

<table>
<thead>
<tr>
<th>Screening Tool</th>
<th>Age</th>
<th>Format (No. of Items)</th>
<th>Time to Complete, min</th>
<th>Reported Sensitivity</th>
<th>Reported Specificity</th>
</tr>
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<tbody>
<tr>
<td>Level 1&lt;sup&gt;a&lt;/sup&gt;</td>
<td></td>
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<tr>
<td>M-CHAT</td>
<td>16–48 mo</td>
<td>Questionnaire completed by parent (23)</td>
<td>5–10</td>
<td>0.85&lt;sup&gt;d&lt;/sup&gt;</td>
<td>0.93&lt;sup&gt;d&lt;/sup&gt;</td>
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<tr>
<td>Level 2</td>
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<tr>
<td>Social Communication Questionnaire (SCQ) (formerly the Autism Screening Questionnaire [ASQ])</td>
<td>≥4 y</td>
<td>Questionnaire completed by parent (40)</td>
<td>5–10</td>
<td>0.85–0.96&lt;sup&gt;e&lt;/sup&gt;</td>
<td>0.67–0.80&lt;sup&gt;e&lt;/sup&gt;</td>
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**Select Screening Tools**

<table>
<thead>
<tr>
<th>Question</th>
<th>YES</th>
<th>NO</th>
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<tbody>
<tr>
<td>1. Does your child enjoy being swung, bounced on your knee, etc.?</td>
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<td>2. Does your child take an interest in other children?</td>
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<td>3. Does your child like climbing on things, such as up stairs?</td>
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<td>4. Does your child enjoy playing peek-a-boo/hide-and-seek?</td>
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<td>5. Does your child ever pretend, for example, to talk on the phone or take care of a doll or pretend other things?</td>
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<td>6. Does your child ever use his/her index finger to point, to ask for something?</td>
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<tr>
<td>7. Does your child ever use his/her index finger to point, to indicate interest in something?</td>
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<td>8. Can your child play properly with small toys (e.g., cars or blocks) without just mouthing, fiddling, or dropping them?</td>
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<tr>
<td>9. Does your child ever bring objects over to you (parent) to show you something?</td>
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<td>10. Does your child look you in the eye for more than a second or two?</td>
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<td>11. Does your child ever seem oversensitive to noise? (e.g., plugging ears)</td>
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<td>12. Does your child smile in response to your face or your smile?</td>
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<td>13. Does your child imitate you? (e.g., you make a face—will your child imitate it?)</td>
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<td>14. Does your child respond to his/her name when you call?</td>
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<td>15. If you point at a toy across the room, does your child look at it?</td>
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<td>16. Does your child walk?</td>
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<td>17. Does your child look at things you are looking at?</td>
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<td>18. Does your child make unusual finger movements near his/her face?</td>
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<td>19. Does your child try to attract your attention to his/her own activity?</td>
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<td>20. Have you ever wondered if your child is deaf?</td>
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<td>21. Does your child understand what people say?</td>
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<td>22. Does your child sometimes stare at nothing or wander with no purpose?</td>
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<tr>
<td>23. Does your child look at your face to check your reaction when faced with something unfamiliar?</td>
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Developmental Screening

Screening tools do not require specialized training and are recommended for use by primary care providers and others.
Developmental Screening

- Act on a positive screening result or when a child demonstrates 2 or more risk factors.
- Do not wait for a definitive diagnosis of an ASD to refer for developmental services.
- Early intervention can be beneficial even if it targets the child's unique deficits.

Do not take a “wait-and-see” approach.
Next Steps: “Search Strategy”

- Studies that should be considered in all children with both an ASD and coexisting GDD/ID.
  - Genetic Microarray (Karyotype)
  - Methyl-CpG-binding protein 2 (MECP2) analysis in females presenting with regression & ASD features (Rett Syndrome)

- Targeted labwork, studies and referrals should be considered when specific clinical findings are identified on an H&P.
Next Steps (Continued)

- Simultaneous referral/testing based on individualized areas of concern *without* waiting for a specialty practice:
  - Audiology (baseline | rule-out hearing issue)
  - Lead screening (developmental delay/pica)
  - Speech Language Pathology (speech delay)
  - Occupational Therapy (fine motor delay)
  - Physical Therapy (gross motor delay)
  - Behavioral Therapy (ABA)

- Early referral can be made to specialty practice for specific ASD Diagnostic Evaluation & Testing.
Overview of Treatment Approaches

- Multimodal, structured interventions have been shown to be effective for many children with ASD and are associated with better outcomes.
  - Educational
  - Behavioral
  - Communication
  - OT/PT (fine or gross motor/ sensory)
  - Pharmacological

- Family support and community resources.
Effective programs are planned, intensive, and individualized interventions within an experienced, interdisciplinary team of providers that includes family involvement to ensure generalization of skills.

Programs share goals of enhancing verbal & nonverbal communication, academic skills, and social, motor, and behavioral capabilities.

The plan should reflect an accurate assessment of the child’s strengths and vulnerabilities with an explicit description of services.
Educational Approaches

- Development of an appropriate Individualized Education Plan (IEP) is *essential* to providing effective service to the child and family.

- Efficacy has been shown for 2 of the structured educational models
  - Early Start Denver Model
  - Treatment and Education of Autism and related Communication handicapped Children (TEACCH) program.
Behavioral Approaches

- Behavioral interventions such as Applied Behavioral Analysis (ABA) are informed by basic and empirically supported learning principles.

- A widely used program for young children is Early Intensive Behavioral Intervention (EIBI).
  - Highly individualized approach.
  - Up to 40 hours/week of 1:1 direct teaching.
  - Uses discrete trials (DTT) to teach simple skills, progressing to more complex skills.

An ABA/DTT Session Demonstration Video
Unit 3: Discrete Trial Teaching (DTT)

Section 1: The Discrete Trial, $S^D$, & Response
Communication Approaches

- Communication is a major focus of intervention with a speech-language pathologist.
- Children who do not yet use words can be helped through the use of alternative communication modalities.
  - Sign language
  - Communication boards
  - Visual supports
  - Picture exchange (eg: PECS)
- For individuals with fluent speech, the focus is on pragmatic language skill development.
Pharmacological Approaches

- Pharmacologic interventions often improve symptoms that interfere with educational and other interventions.

- Help individuals remain in less restrictive environments.

- Frequent targets for pharmacologic intervention include:
  - Aggression and/or self-injurious behavior
  - Hyperactivity and/or inattention
  - Compulsive, repetitive or stereotypic behaviors
  - Anxiety, Depression, Psychosis
  - Sleep disturbance
Summary

- ASD is the 5th most prevalent pediatric mental health disorder in the United States.
- ASDs significantly impact families, communities, and society.
- ASDs are complex, multi-determined neurodevelopmental disorders.
- ASDs can be reliably diagnosed at a young age.
- Need for universal developmental screening.
- Early diagnosis & treatment improves outcomes.
- Treatment is necessarily multi-modal.
Thank You

Questions