Assessment and Management of Autism Spectrum Disorder in the Primary Care Setting

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Disclosures

• Relevant Financial Disclosures

I have no financial relationships or affiliations related to this presentation to disclose.
Disclosures

• Non-FDA Approved Uses
  o Brief discussion of off-label use of pharmacotherapeutics in specialty care practice and implications for primary care
  
  o Examples: Selective Serotonin Reuptake Inhibitors (SSRIs); stimulants; alpha-2 adrenergic agonists; atypical neuroleptics
Autism Spectrum Disorder: Prevalence

• 5th most prevalent pediatric mental health disorder in the United States ¹

• Rapidly increasing prevalence:
  o 1943: RARE – 2-4 per 10,000 children²
  o 2006: Approximately 1 in 110 children³
  o 2010: Approximately 1 in 68 children¹
Autism Spectrum Disorder: Prevalence

• Disproportionately impacts **boys over girls** at a rate of almost 5:1 ³
  - 1 in 42 **boys** in the United States ³
  - Studies suggest comparable or higher prevalence rates in other parts of the world ³,⁴
Autism Spectrum Disorder: Etiology

- 10% of cases have identifiable genetic and chromosomal etiologies (Fragile X Syndrome, tuberous sclerosis, metabolic syndromes) \(^5\)

- Majority of cases (90%) are **idiopathic** with potentially multiple genetic determinants and environmental contributing factors \(^5\)
Autism Spectrum Disorder: Defined

- Complex and potentially multiple etiologies
  - The Autisms

- ASD is a group of disorders currently diagnosed clinically by core symptoms set forth in the Diagnostic and Statistical Manual of Mental Disorders (DSM V)
Autism Spectrum Disorder: DX Criteria – The Long Form

- Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria) 7

- 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):
  - 1. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back-and-forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
Autism Spectrum Disorder: DX Criteria – The Long Form

- Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7
  - 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.
Autism Spectrum Disorder: DX Criteria – The Long Form

- Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7
  
  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.

- Severity is based on social communication impairments and restricted repetitive patterns of behavior.
Autism Spectrum Disorder: DX Criteria – The Long Form

• Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7

• 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):
  ○ 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).

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Autism Spectrum Disorder: DX Criteria – The Long Form

• Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7

  o 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).
  o 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).
Autism Spectrum Disorder: DX Criteria – The Long Form

- Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7
  
  - 4. Hyper- or hypo-reactivity 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).

- Severity is based on social communication impairments and restricted, repetitive patterns of behavior.
Autism Spectrum Disorder: DX Criteria – The Long Form

• Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7

• C. Symptoms must be present in the early developmental period (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life).

• D. Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning.
Autism Spectrum Disorder: DX Criteria – The Long Form

- Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7

- 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.
Autism Spectrum Disorder: DX Criteria – The Long Form

• Diagnostic and Statistical Manual of Mental Disorders (DSM V Criteria Continued) 7

• Note: Individuals with a well-established DSM-IV diagnosis of autistic disorder, Asperger’s disorder, or pervasive developmental disorder not otherwise specified should be given the diagnosis of autism spectrum disorder. Individuals who have marked deficits in social communication, but whose symptoms do not otherwise meet criteria for autism spectrum disorder, should be evaluated for social (pragmatic) communication disorder.
Autism Spectrum Disorder: Diagnosis Criteria Summary

- Communication and social interaction deficits
- Restricted repetitive language and behaviors
- Inflexibility and rigidity
- Sensorimotor dysregulation (hypo and hyper-reactivity)
- Impact: Mild to Severe
Why Does This Matter to Primary Care Nurse Practitioners?

• Per the Council of Pediatric Subspecialties:
  - **HUGE SHORTAGE** of specialty providers in pediatric mental health across the U.S.
  - Less than 7,500 practicing child and adolescent psychiatrists and less than 600 developmental behavioral pediatricians
  - Only 20% of children with emotional/behavioral problems receiving any mental health treatment 8,9
Why Does This Matter to Primary Care Nurse Practitioners?

• Shortages of specialty providers and shortages in State funding can mean delays of months to over a year for access.

• With no current curative interventions, early and intensive intervention has been found to be the most effective strategy for maximizing function in ASD.
Role of Nurse Practitioners in ASD Assessment and Management

• Early identification and timely referral
  o For Evaluation/Diagnosis, Audiology, PT, OT, Speech, Behavioral

• Assessment and management of common problems and comorbidities seen in ASD
  o Alteration in Nutrition, GI/Toileting, Sleep Disturbance

• Co-management of medications prescribed in specialty practice
Early Identification of ASD

- Autism Spectrum Disorder often presents **prior to the age of 2 years** and initial symptoms may be apparent at birth\(^{10}\)

- **Listen to parents:** Their concern is a good predictor of atypical development prior to 24 months\(^{10,11}\)

- Better outcomes are linked to **more intensive behavioral regimens started at a younger age** (prior to 2-3 years of age) but there is no point where not beneficial\(^{11}\)
Early Identification of ASD: RED FLAGS

RECOGNIZE THE SYMPTOMS:

- Not respond consistently to their name by 12 months
- Not point at objects to show interest by 14 months
- Not play "pretend" games by 18 months (feed a baby)
- Avoid eye contact and want to be alone
- Have delayed speech and language skills
- Repeat words or phrases over and over (echolalia)
- Have trouble understanding other people's feelings or talking about their own feelings
Early Identification of ASD: RED FLAGS

RECOGNIZE THE SYMPTOMS:¹⁰

- Give unrelated answers to questions
- Get upset by minor changes and transitions
- Have obsessive interests
- Flap their hands, rock their body, or spin in circles
- Have unusual reactions to the way things sound, smell, taste, look, or feel
- Demonstrate repetitive or atypical play (lining up long rows of cars, patting toys)
Early Identification of Children at Risk for ASD: Screening

American Academy of Pediatricians:\textsuperscript{12}

- Screen \textbf{ALL} children for developmental delay:
  - At 9 months
  - At 18 months
  - At 24 months
  - At 30 months
  - Surveillance throughout school age years
  - \textbf{HIGH RISK:} sibling or parent with ASD, preterm, low birth weight
Early Identification of Children at Risk for ASD: Screening Tools\textsuperscript{13, 14}

**Ages and Stages Questionnaires (ASQ):**
- General developmental screening tool
- Parent-completed questionnaire
- P/F screen per domain: communication, gross motor, fine motor, problem-solving, and personal adaptive skills.

**Communication and Symbolic Behavior Scales (SCBS):**
- Standardized tool for screening communication and symbolic ability up to 24 months
- 1 page parent completed tool
Early Identification of Children at Risk for ASD: Screening Tools 13, 14

**Parents’ Evaluation of Developmental Status (PEDS):**
- General developmental screening tool
- Parent interview form
- Screens for developmental and behavioral problems
- Single response form used for all ages--may be useful as a surveillance tool

**BRIGANCE® Early Childhood Screens:**
- Teacher/parent rating forms
- Assess: language, motor, self-help, social-emotional, cognitive domains
Early Identification of Children at Risk for ASD: Screening Tools 13, 14

**Modified Checklist for Autism in Toddlers (MCHAT)**
- Parent-completed questionnaire
- Designed to identify children at risk for autism in the general population (valid from 18 months)

**Screening Tool for Autism in Toddlers and Young Children (STAT):**
- Interactive screening tool designed for children with suspected developmental concerns
- 12 activities assessing play, communication, and imitation
- Takes 20 minutes to administer
Early Identification of Children at Risk for ASD: Screening Tools

Screening tools no longer recommended by the American Academy of Neurology and the Child Neurology Society for primary care screening:

- Denver-II (DDST-II)
- Revised Denver Pre-Screening Developmental Questionnaire (R-DPDQ)
- Decreased specificity and sensitivity for ASD
Early Identification of Children at Risk for ASD: Screening Tools $^{13, 14}$

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

- Early referral can be made to **specialty practice** for additional **diagnostic** testing:
  - ADOS-G, ADI-R, CARS-2, GARS-2
Early Identification of Children at Risk for ASD: Screening Tools \textsuperscript{13, 14}

Simultaneous referral/testing: Based on individualized areas of concern \textit{without} waiting for specialty practice:

- Audiology (baseline/rule out hearing impaired)
- Lead screening (developmental delay/pica)
- Speech Language Pathology (speech delay)
- Occupational Therapy (fine motor delay)
- Physical Therapy (gross motor delay)
- Behavioral Therapy (ABA) (behavior concerns)
Assessment and Management of Common Concerns in ASD

- Feeding/Alteration in Nutrition
- GI Pain, Elimination, Toileting
- Sleep Disturbance
Feeding/Nutrition Issues in ASD

• Atypical feeding: identified in ASD as early as 1943

• Clinical symptoms of ASD that contribute:
  o sensory issues, rigidity, social interaction deficits

• Feeding issues frequently identified by parents:
  o Food selectivity (limited intake)
  o Food aversions (smell, texture, color, temp, brand)
  o Food refusal (preferred carbs/snacks)
  o Disruptive mealtime behaviors\textsuperscript{16}
Feeding/Nutrition Issues in ASD

Long term risks of unhealthy feeding patterns in ASD:

- **Obesity:** Recent National Health Interview Survey (2008-2010) of over 9,500 adolescents found obesity highest in autism subgroup, with 2x higher prevalence than same-age neurotypical peers\(^{17}\)

- **Nutritional deficiency:** Meta-analysis using cumulative sample of 881 children with ASD showed:
  - feeding problems were 5x more likely in children with ASD than neurotypical peers
  - children with ASD had significantly lower calcium and protein intake compared to neurotypical peers\(^{18}\)
Feeding/Nutrition Issues in ASD: Assessment

• APN assessment for this population needs to include:  
  
  o **Comprehensive history**—eating behaviors, diet (?restrictive: GFCF/SF, others), pica, supplement use, allergies, medications, past medical conditions, **pain and impaired swallow**
    • Use of the Brief Autism Mealtime Behavior Inventory (BAMBI) and Food Inventories
  
  o **Complete physical and neurological exam**—assess changes—pain, functionality, anemia, constipation, GI
Feeding/Nutrition Issues in ASD: Management

- **Rule out physiologic/neurologic/allergic conditions:**
  - impaired chewing/swallowing, reflux, food allergy, obstruction
- **Consider labs for suspected deficiency/MVI use:**
  - CBC, calcium, protein
- **Track:**
  - height, weight, growth velocity, and eating patterns
- **Counsel families on nutrition/mealtime behaviors:**
  - establish routine, consistent expectations, repeated reintroduction of non-preferred foods, pairing preferred with non-preferred foods, establishing behavioral limits, stretch dietary repertoire
- **Refer** to appropriate resources
  - Nutrition, Behavioral, GI\textsuperscript{16,19}
Common GI Problems, Elimination, and Toileting Issues in ASD

Increased behavioral problems in ASD may have roots in physiologic causes, especially GI:

- Withholding patterns
- Abdominal pain
- Constipation/Encopresis
- Diarrhea
- Gastroesophageal reflux
- Food allergies

GI dysfunction may occur in 9-70% of children with ASD\textsuperscript{16,20}
Common GI Problems, Elimination, and Toileting Issues in ASD

- Evaluation of GI problems may be complicated by communication deficits and inability to localize pain symptoms

- Presentation may be atypical
  - Change in behavior may be the only clue to underlying pathology\textsuperscript{16,20}
Common GI Problems: Withholding Behavior in ASD

Withholding behavior can contribute to abdominal pain, constipation, encopresis, and urinary tract problems. Common triggers include:

- Unfamiliar environment
- Aversive stimuli
- Change associated with sensory issues (from diaper to open air)
- Change in routine
- Pain secondary to constipation
Common GI Problems: Abdominal Pain in ASD

**Abdominal pain behaviors** in children with ASD can present typically or as:

**Vocalizations:**

- Frequent throat clearing, swallowing, tics, spitting
- Sobbing for no reason, sighing, whining, moaning, groaning
- Delayed echolalia with reference to stomach or tummy
- Direct verbalizations of pain with pointing ("ow", "ouch", "bad")
Common GI Problems: Abdominal Pain in ASD

**Abdominal pain behaviors** in children with ASD can present typically or as:

**Motor behaviors:**

- Grimacing, wincing, teeth gritting
- Constant eating/drinking (“grazing”)
- Mouthing behavior: chewing shirts, clothes, toys, pica
- Applying pressure to abdomen
- Unusual posturing (jaw thrust, back arching, etc.)
- Aggression, self-injury

\(^{20}\)
Common GI Problems: Abdominal Pain in ASD

Abdominal pain behaviors in children with ASD can present typically or as:

Changes in Overall State:

• Increased difficulty sleeping
• Increased irritability
• Increased agitation
• Food aversion
• Noncompliance with demands that generally were done with appropriate response (increased oppositional behavior)
Common GI Problems: Constipation in ASD

**Constipation** is the *most common GI complaint* in children with ASD and is exacerbated by diet. Consider when:

- Straining or infrequent stool
- New onset of diarrhea or fecal incontinence (*encopresis*)
- New self-injurious behavior, tantrums, aggression, oppositional behavior
- New problem behaviors with meals
- Abdominal discomfort, bloating, flatulence\textsuperscript{20}
Common GI Problems in ASD: Assessment

ALL potential GI symptoms warrant thorough evaluation:\textsuperscript{16,20}

- **Comprehensive history**—eating behaviors, change in general behavior, diet (restrictive: GFCF/SF, others), pica, supplement use, allergies, medications, past medical conditions, pain, impaired swallow, nausea, vomiting, bowel, bladder patterns, toileting and sleep behaviors.

- **Complete physical and neurological exam**—assess changes—pain, functionality, wt change, GI signs, impaction, rectal bleeding.
Common GI Problems: Management by Symptoms

Consider for DD and management:

- **Constipation**: Hard infrequent stool, straining behavior, prolonged BR time, perceived abdominal discomfort, self injury, tantrums, oppositional behavior, flatulence, bloating

- **RED FLAGS**: fever, distension, n/v, anorexia, wt loss/gain

  - Labs: TSH, T4, calcium, lead, celiac screen; trial of PEG; diet enhancements (fruit, fiber, fluids); bowel training; abdominal radiograph

  - Refer to appropriate resources—GI, Nutrition, Behavioral \(^{16,20,21}\)
Common GI Problems: Management by Symptoms

Consider for DD and management:

- **Diarrhea (chronic > 3 stools/d x2wks):**
  Overflow (encopresis), malabsorption, maldigestion, ulcerative colitis, food allergy

  • Allergy panel, celiac panel, stool for occult blood, enteric pathogens/parasites (Giardia, Cryptosporidium, C. difficile); trial of PEG if suspected overflow diarrhea; lactose breath test; EGD or colonoscopy

  • Refer to appropriate resources—GI, Nutrition, Behavioral\textsuperscript{16,20}
Common GI Problems: Management by Symptoms

Consider for DD and management:

- **GERD**: Sleep disturbance, self-injurious/aggressive/oppositional behavior, perceived abdominal discomfort, food aversion. Consider gastritis and intestinal inflammation.

  - Trial of PPI; may require pH probe, EGD
  
  - **Refer** to appropriate resources—GI, Nutrition, Behavioral $^{16,20}$
Common GI Problems: Toileting

One of the greatest sources of stress to parents regardless of ASD
Common GI Problems: Toileting

When counseling parents, be aware:

• Most children with ASD learn to urinate and have BMs in the toilet **later** than other children (6+ years is not unusual)

• Children with ASD may take years to achieve bladder/bowel control

• Each child is different--understand the reasons why each child might be having difficulty22
Common GI Problems: Toileting

Toileting Barriers in ASD:

- Physical or medical issues (neuro, GI)
- Language deficits in expressing need
- Dressing/fine motor coordination
- Fear (flushing noise, sitting on toilet)
- Body cues (inability to sense need to go)
- Change in routine (from diaper to toilet)
- Change in environment (public toilet)
- Aversive sensory stimuli$^{23}$
Common GI Problems: Toileting

Management Strategies:

• Rule out medical reasons for toileting issues (obstruction, neurological)

• Reassure parents--keep trying!!!!

• Direct parents to appropriate resources: — Autism Speaks/ATN

• **Refer** for OT or Behavioral Assistance²³
Sleep Disturbance in ASD

- Problems with *sleep latency* and *night waking* seen in 50-80% of school aged population with ASD

- **Chronic insomnia** may be *10x higher* in ASD

- **Sleep problems may increase** rather than decrease as children grow older\textsuperscript{24}
Sleep Disturbance in ASD

- Overlap of **neurobiological factors** seen in both sleep disturbance and ASD

- Abnormalities in:
  - **GABA** (an inhibitory neurotransmitter)
  - **Melatonin** (a brain hormone associated with circadian sleep-wake cycles)\(^\text{24}\)
Sleep Disturbance in ASD

Sleep disturbance is linked to:

• Worsened daytime behaviors
  – Anxiety, aggression, tantrums

• Increased core symptoms
  – Stereotyped behavior, communication deficit, ASD severity

• Disrupted family function and caregiver stress
Sleep Disturbance in ASD: Assessment

• Ask parents about:
  o Bedtime/bedtime rituals (parents needed?)
  o Sleeping arrangements
  o Patterns: latency, duration, night waking
  o Medications/supplements for sleep
  o Breathing problems, *snoring*, HEENT issues
  o Daytime sleepiness/increased behaviors

• Assessment tools: Children’s Sleep Habits Questionnaire (CSHQ)\textsuperscript{26}
Sleep Disturbance in ASD: Management

- **Rule out physiologic causes:**
  - Tonsils/adenoids, ears/sinus/nasal, OSA, GERD

- **Sleep Hygiene:**
  - Routine, limit stimuli before bed (TV, computer, food, fluids, caffeine)
  - Get out of bed after 15-20 min; OOB relaxing activity
  - Consider noise disturbance, use of white noise

- **Nonbehavioral interventions:**
  - Consider melatonin (no standard dose: adult dose is 3.5 mg at hs – varies per individual, **dark** is important)
  - Specialty provider: centrally acting alpha agonists
  - Diphenhydramine not generally effective/rebound

- **Refer** to appropriate resources
  - ENT/GI, Sleep Study, Behavioral \(^{24}\)
Co-Management of Medications Used in Specialty Practice for ASD

- Limited FDA approved pharmacological interventions specifically for ASD
  - There are currently **TWO**

- Both are atypical neuroleptics with SE profiles that impact primary care:
  - aripiprazole (for irritability)
  - risperidone (for irritability)

- NPs: **specialty care prescribed** with collaborative management⁹
Co-Management of Medications Used in Specialty Practice for ASD

○ Off-label medications used in specialty care:
  • **SSRIs** (anxiety, depression, obsessive-compulsive behaviors)
  • **Stimulants** (hyperactivity, inattention, impulsivity)
  • **Centrally acting alpha-2 adrenergic agonists** (clonidine, guanfacine) (tic reduction, hyperactivity, impulsivity)
  • **Nonstimulant alternatives** (atomoxetine) (hyperactivity, impulsivity, inattention)
Medications in Specialty Practice for ASD: Primary Care Concerns

- **Atypical Neuroleptics:** Consider risk for metabolic syndrome—Ht, Wt, BMI, truncal obesity, BP, P; Labs: CBC, CMP, lipids, prolactin levels, HgbA1C

- **SSRIs:** Consider black box warnings for SI/SA in peds; may cause disinhibition, HA, GI SEs, wt gain, insomnia, serotonin syndrome (small risk); **taper** to avoid withdrawal sx⁹
Medications in Specialty Practice for ASD: Primary Care Concerns

- **Stimulants**: Consider appetite suppression, weight loss, mild growth suppression—Ht, Wt, BMI, BP, P, tics; consider cardiac evaluation for (+) family/personal hx

- **Centrally acting alpha-2 adrenergic agonists**: Consider sedation, hypotension, bradycardia, constipation, drymouth, increased appetite—Wt, BMI, BP, P
Medications in Specialty Practice for ASD: Primary Care Concerns

- Nonstimulant alternatives (SNRIs): (atomoxetine) SEs similar to stimulants (excluding tics); SEs may also include cough, dizziness, hepatotoxicity, and small risk of SI/SA—Ht, Wt, BMI, BP, P, LFTs

- Collaborative care is critical to our ability to manage these patients in primary care!
Last But Not Least—Support Families and Caregivers

- Knowing available community resources is essential (delays and wait times matter)
- Caregiver strain is HUGE in this population
- Families are struggling to find support and guidance—we often find it from other parents
- NPs are ideally suited to help in key areas primary care management, educational support and access to community resources
Community Resources for ASD

- Regional Center of Orange County (RCOC)
- UCI Center for Autism and Neurodevelopmental Disorders
- CHOC Early Developmental Assessment Center
- Help Me Grow
- Early Childhood Education Programs (IUSD, SVUSD, Costa Mesa, others)
- United Cerebral Palsy (UCP), Irvine
Conclusion: ASD and Primary Care

• Primary care NPs are critical to:
  o Early identification/referral
  o Management of key areas of concern
    • Nutrition
    • GI symptoms
    • Toileting
    • Sleep
  o Collaborative management
  o Supporting families
Conclusion: ASD and Primary Care

WE NEED YOUR HELP!
Conclusion: ASD and Primary Care

THANK YOU!
References

References


References