Autism Spectrum Disorder

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Learning Objectives

- Understand the core features of autism and how they present throughout the life course
- Learn the impact of the medical vs social model of disability on autistic individuals
- Recognize common co-occurring diagnoses
- Increase comfort in recognition and management of physical, mental, and behavioral concerns that may occur in autistic individuals

What is autism?[1]

Biologically based

Diagnosis based on

- Difference in social communication and interaction
- Repetitive or restrictive patterns of behavior, interests, or activities

Signs and Symptoms are present in early childhood

- May not become obvious until later in childhood, or adulthood, when demands increase

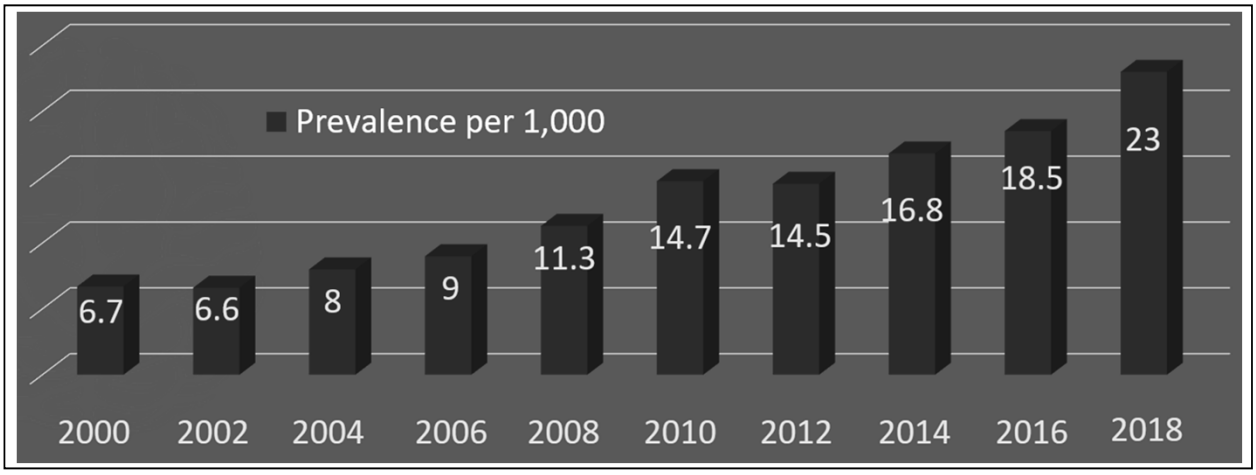
Etiology of autism?[2-5]

- Genetic factors
  - 40-90% heritability rate
  - Most autistic people do not have monogenic findings
  - Reasonable to offer genetic testing to any individual diagnosed as autistic.
- Prenatal/Perinatal exposures?
- No evidence to support vaccines causing autism

Prevalence of autism?

Most recent CDC estimates:
- 1 in 44 children
- 2.2% of adults

Prevalence of Autism Spectrum Disorder
Data from ADDM Network 2000-2018 [6]



Medical vs Social Model of Disability

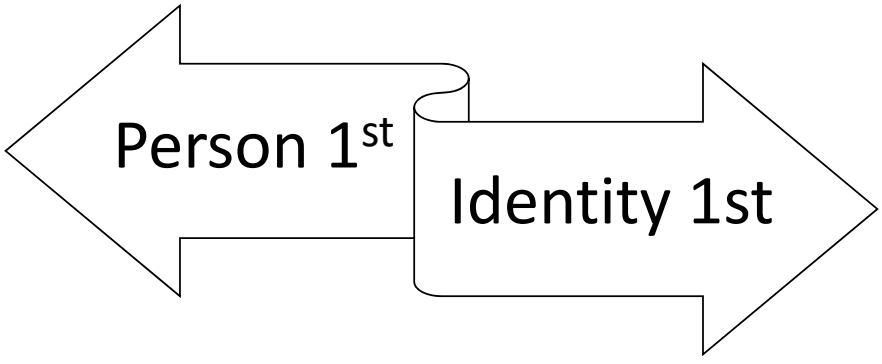
Medical Model

- Focused on the individual and
  - Their impairment
  - Remediation of skills so that an individual can progress along a typical developmental pathway
- The source of words like “disorder” and “deficit”
- The search for a cure for autism

Social Model

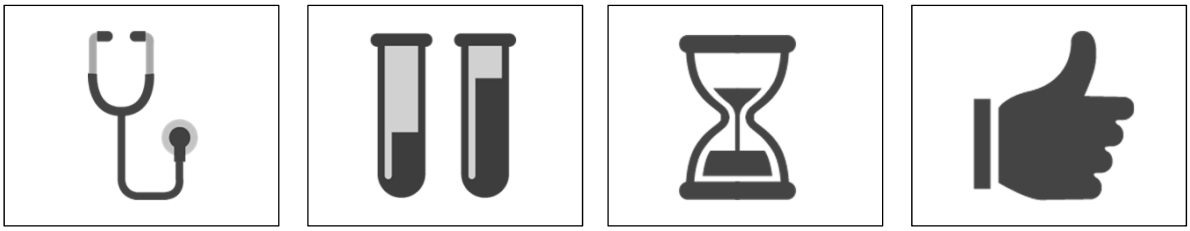
- Views the disability as a result of:
  - Environmental barriers
  - Societal barriers
- Looks at not just impairment, but also strengths and abilities
- Neurodiversity movement

Language of autism[7]



Screening for autism

- MCHAT-R/F[8]: age 16-30 months
  - Sensitivity 0.83
  - Specificity 0.94
- ASSQ[9]: age 7-16 years
  - Sensitivity 91%
  - Specificity 86%
- AQ[10]: adults
  - Sensitivity 79%
  - Specificity 98%



Diagnosis of Autism

Comprehensive Evaluation

- History and physical
- Diagnostic tools: ADOS, ADI-R, CARS-2, other

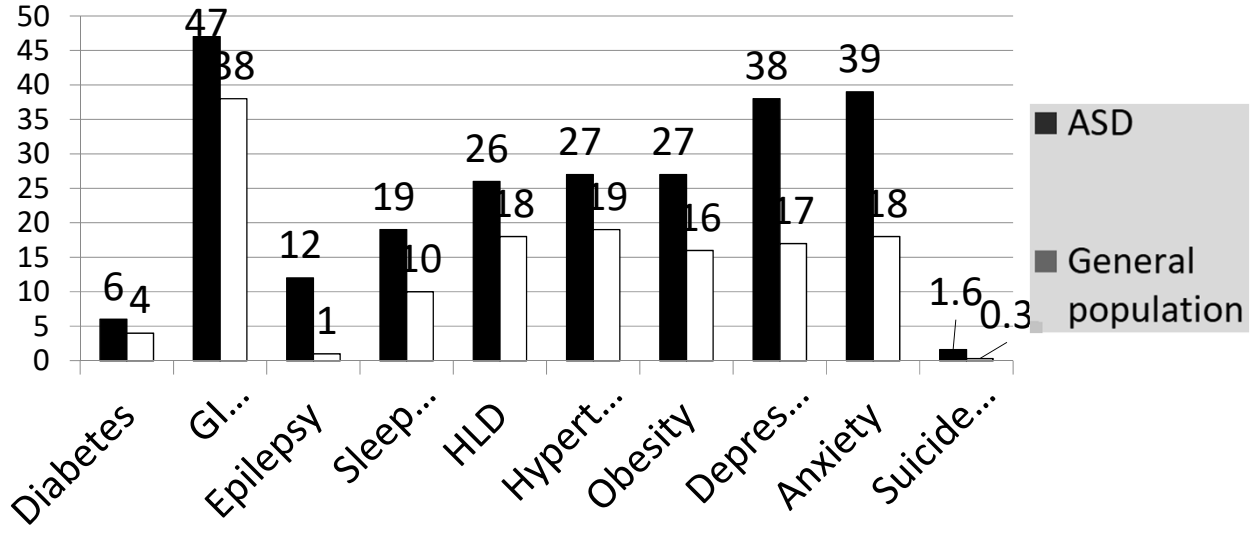
No need to wait for diagnosis to refer for intervention

The Role of Primary Care for Autistic Patients

1	2	3	4
Avoid diagnostic overshadowing	Age-appropriate health care	Accommodate for individual needs	Refer to appropriate resources

Physical Health and Autism

Percent of population with medical conditions[11]



Preventative Healthcare and Age-appropriate care

- Age-appropriate screenings
  - Cancer screenings
  - Labs
  - Screening for substance abuse
  - STI screening
- Vaccinations
- Other

Providing appropriate accommodations

Clinical settings with systems focused on accommodations can result in[12-15]:

- Higher likelihood of preventative healthcare services
- Higher levels of satisfaction of care
- Fewer unmet healthcare needs
- Higher continuity of care
- Lower expenditures for mental health admissions
- Fewer inpatient hospitalizations and ED visits

Referring to appropriate supports and resources

- Therapies: ST, OT, PT, Behavioral Health, ABA, other
  - Early intervention, School-based, Private, etc
- Physician specialists
- Social supports:
  - Board of developmental disabilities
  - School-based
  - Vocational supports
- Key ages to think about
  - Under 3 years – early intervention
  - 18 years – supported decision-making
  - 22 years – end of school-based special-education supports
  - 26 years – change from parent-based insurance

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Behavioral and Psychiatric Manifestations of Autism Spectrum Disorder

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Behavioral Manifestations

Restrictive and Repetitive Behaviors (RRBs)

- Essential feature of ASD
  - Repetition
  - Inflexibility
  - Invariance
  - Inappropriateness and lack of obvious function/purpose
  - Restricted/fixated interests

Repetitive motor behaviors – stereotypies, rocking, hand flapping, moving fingers in front of eyes, spinning toys, flipping light switches repeatedly

Ritualistic behaviors – insistence on sameness, resistance to change, repetitive language, limited interests, eating same foods, watching same movie (or even parts) repeatedly, resistance to change in environment

Behavioral Manifestations

Restrictive and Repetitive Behaviors (RRBs)

Beneficial vs Impairing

- Pt with fixated interest in weather – pursuing career in meteorology; delays sleep to track storms

Functional impairment

- school performance
- learning essential life skills
- social relationships
- problem behaviors to discourage change/interruption

Behavioral Manifestations

Challenging (Problem) Behaviors

Estimated 50+% of individuals engage in at least one
They are hard to address, and can significantly affect daily life

- Aggression
  - scratching, biting, kicking
  - property destruction
- Self-injurious behavior
  - scratching, hair pulling, headbanging
- Tantrums
- Noncompliance

Behavioral Manifestations

Challenging (Problem) Behaviors

Operant Conditioning

Antecedent (stimulus) → Behavior → Consequence(s)

Antecedents can trigger, consequences can reinforce behavior

- Escape a demand
- Denial
- Punishment
- Medical

'Setting events'

- internal – biological
- external – environmental

Common internal

- Allergies
- Anxiety
- Constipation/GERD
- Migraines
- Sleep disorders
- Pain

Behavioral Manifestations

Challenging (Problem) Behaviors

Internal Sensory Perception (Interoception)

Mediated by anterior insula and ventromedial prefrontal cortex

Studies suggest impaired function in autism

- Hyperresponsive
- Hyporesponsive

Communication Difficulty

Limited or inability to express wants and needs → frustration → CB

Challenging behaviors are a primary reason individuals come to psychiatric attention

Psychiatric Manifestations

Co-Occurring Psychiatric Conditions

Anxiety Disorders - 40-80%
ADHD – 30-50%
Obsessive Compulsive Disorder - ~17%
Depression – 10-70%
Bipolar Disorder – 5-8%
Psychosis - ~35%
Catatonia – 12-18%

Psychiatric Manifestations

Assessment of Psychiatric Conditions

Features of ASD can overlap with different psychiatric conditions

- Determine patient's baseline
- Consider developmental level
- Rule out medical conditions
  - hypothyroidism and OSA for depression
- Consider genetics
  - William's Syndrome – anxiety
  - 22q11 Deletion Syndrome - psychosis

Anxiety Disorders

Risk factors: social skills, sensory sensitivity, rigidity

Associated with challenging behaviors

Specific Phobia

- Sensory driven
- Noise, needles, large crowds
- Can present as avoidance behaviors

Anxiety Disorders

Generalized Anxiety

- Atypical features
  - preoccupations
  - schedule/change
- Can present as repetitive questions, reassurance seeking about specific events/worries
- Some have difficulty vocalizing - fidgeting, pacing, hand-wringing

Social Anxiety

- Distinguish lack of interest in socialization from avoidance due to fear of embarrassment/judgment

ADHD

Impairments in attention, hyperactivity, and impulsivity

Overlapping features:

- Inattention
- Executive dysfunction
- Social/communication deficits
- Impulsivity
- Restlessness
- Hyperactivity

DSM-IV vs DSM-5

ADHD + ASD

- Increased psychosocial difficulty
- Lower quality of life

Assessment

- Distractibility related to fixated interest, sensory seeking behavior
- Lack of focus due to disinterest rather than concentration
- Excessive talking/interruption – specific interest?

Obsessive Compulsive Disorder

Obsession - Recurrent and persistent thoughts, urges, or images that are experienced… as intrusive and unwanted, and… cause marked anxiety or distress.

Compulsion - Repetitive behaviors (e.g., hand washing, ordering, checking) or mental acts (e.g., praying, counting, repeating words silently) that the individual feels driven to perform in response to an obsession or according to rules that must be applied rigidly.

Overlapping features - RRBs

Key differences:

- OC – distressing, bothersome, unwanted
- ASD – preferred or comforting; consider stereotypies, ritualistic behaviors

Psychotic Disorders

Hallucinations, delusions, disorganized speech, disorganized behaviors

Overlapping features:

- Language pragmatics - disorganized speech
- Under stress – can appear disorganized
- Idiosyncratic/Scripted language – disorganized/delusional
- Restricted interests – delusional
- Theory of Mind – distrust/paranoid
- Self-dialogue – hallucinations

Assessment:

- Developmental history
- Psychotic symptoms usually develop in adolescence or adulthood
- Features above which have been present for years since childhood more likely d/t ASD
- Key is **differentiating baseline and assessing change** in function

Catatonia

Complex neuropsychiatric behavioral syndrome characterized by abnormal movement, communication, behaviors and withdrawal

- In neurotypical population associated with mental and medical illness
- In ASD can develop in adolescence/early adulthood

Presentation

- Immobility/Stupor
- Mutism
- Staring
- Posturing
- Grimacing
- Echopraxia/echolalia
- Stereotypies
- Withdrawal
- Verbigeration

Catatonia

Overlapping features:

- Echolalia
- Mutism
- Stereotypic movements

Assessment:

- Developmental history, baseline behaviors and assessing for change
- Loss of skills (self care, toileting, etc) – needing prompts or guidance on things previously mastered
- Reduced speech
- Difficulty starting tasks
- Getting "stuck"
- Repetitive behaviors – if present at baseline, have they increased?

Management of Behavioral and Psychiatric Concerns

Behavioral Interventions

Mainstay of treatment

Challenging Behaviors

- Medical exam – acute or chronic illness, pain, etc.
- Psychiatric exam

Applied Behavioral Analysis (ABA)

Most well-known therapy for children with autism

Features

- Social skills, communication, self-care, learning, etc. (individualized)
- School, home, community
- Several hours per week
- Antecedents and Consequences (Functional Behavioral Assessment)
- Goal – to be independent as much as possible; reduce challenging behaviors

Consistent use CAN improve behaviors/skills

Controversy

- Past iteration – positive and negative reinforcement (punishment for 'failure')
- Too intense for young children
- Neurodiversity (autism is a normal variation; "not-autistic")

Psychotherapies

Cognitive Behavioral Therapy

Thoughts, emotions, and behaviors

Helpful for co-occurring depression, anxiety

Adaptations

- Written, visual information
- **Behavior >> Cognitive strategies**
- Concrete language
- **Psychoeducation about emotions**

Dialectic Behavioral Therapy

Thoughts, emotions, and behaviors + mindfulness, emotion regulation, and distress tolerance

Helpful for suicidal ideation, self harm, destructive behaviors, emotional dysregulation

Pharmacologic Treatments

There are **NO** medications for the core symptoms of autism

Focus on treating co-occurring conditions

Medication selection follows similar algorithm as NT population

- Higher sensitivity to effects
- Increased likelihood of adverse effects
- Adjust titration schedule

Years of research, though inconsistent results in efficacy in treatment studies

- Varied etiology of “autism”

Pharmacologic Treatments

Serotonergic Agents

Regulate serotonin
- GI
- CV
- CNS

Theory: serotonin dysregulation → repetitive behaviors, anxiety, irritability, etc.

SSRI >>> SNRI, TCA

Atypical Antipsychotics

2 FDA approved for irritability associated with autism
- Risperidone
- Aripiprazole

Dopamine, serotonin, alpha-adrenergic, histaminergic receptors in CNS

Regular monitoring for side effects

Periodically re-evaluate need to continue treatment

Pharmacologic Treatments

Stimulants

Usually 1st line for co-occurring ADHD

PMH, FMH, PE (CV)

Side effects: decreased appetite, HTN, weight loss, sleep disruption, headache

Baseline sleep issues not necessarily predictive of stimulant related sleep issues

AMP – slightly more effective

MPH – usually better tolerated

Alpha-2-adrenergic Agonists

Clonidine and Guanfacine

Nonstimulant treatment for ADHD

Less effective than stimulants

Can be helpful with co-occurring sleep issues

Small study showing clonidine has positive effects
- Irritability
- Stereotypies
- Hyperactivity
- Hyperarousal

Social Supports

Board of Developmental Disabilities
School Based

Behavioral Support
Daily Living Skills
Social Skills Training/Groups

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