Missed Diagnosis or Misdiagnosis?

Population Considerations and Clinical Comorbidities Contributing to Delayed Diagnosis of Autism Spectrum Disorder

Laura Rowley, PhD
Utah Center for Evidence Based Treatment

Intro- Why I am Here

- Assessment and Testing Program leader at UCEBT
- Doctorate in Clinical Psychology at Wayne State University in Detroit, MI
- Internship and Postdoctoral Fellowship at Primary Children's Hospital
- Big sister of two siblings on the Autism Spectrum
- Evaluating for autism is incredibly complex!

Who This talk is for

You are a person who assesses for Autism Spectrum Disorder or works clinically with that population

You are an educator or healthcare provider

You know someone on the Autism Spectrum

You want to learn more about Autism

The Spectrum

- DSM-5 Criteria outlin... ...ains of deficits in Autism Spectrum Disorder (ASD...
- Social co...
  - Socia...
  - Nonv...
  - Develop...
- Restricted and re... ...vior
  - Stereotyped/repetitive movements
  - Insistence on sameness, routines, rituals
  - Highly restricted or fixated patterns of interest
  - Low or high reactivity to sensory input and/or unusual interest in sensory aspects of environment

ASD requires BOTH the ABSENCE of typical social behaviors and the PRESENCE of atypical behaviors

Levels of Support

- Severity of ASD falls in one of three levels reflecting the amount of support needed
  - Level 1 requires the least and Level 3 requires the most support
- Terms like "high-functioning" and "low-functioning" are inaccurate and can be harmful
- This talk will focus on Level 1
  - These are the kids most likely to be missed
  - 44% of individuals with ASD have average to high average intellectual abilities

CDC, March 2020- based on a sample of children age 8

Autism In the Population

- 1 in 54 children diagnosed
- Occurs in all ethnic/racial groups
- Prevalence is increasing across all groups…
  - Are we overdiagnosing or better at recognizing?
- …but we are still not capturing some
  - 1:4 or 1:3 ratio of girls to boys receiving diagnosis
  - ASD diagnosis among white children exceeded black children by 7% and Hispanic children by 22% in 2014

Looms, et al., 2017 ;Maenner et al., 2016

What people often think ASD is…

Atypical; Netflix

Criminal Minds; CBS

The Good Doctor;
ABC

The Big Bang Theory;
CBS

ASD in Individuals Socialized as Girls

The "Female Autism Phenotype"

- Less repetitive behaviors or restrictive interests
- Special interests may be socially acceptable
- Impairment in relationships are qualitative, rather than a complete lack of forming friendships
- Deficits don't often show up until later childhood or adolescence
- Demonstrate better emotion recognition and empathetic responding
- More socially motivated

Hendrix, 2015; Leedham et al., 2020; Nichols, 2009; Wilson et. Al, 2016

This looks like…

- Having friendships in childhood and not keeping up with demands of complex relationships in adolescence
- Not recognizing signs of relational aggression or not being aware of when others are taking advantage
- Appearing immature for their age and fears of growing up
- Watching on the sidelines, copying and practicing, expending a significant effort on appearing "normal"
- Sensory overload leading to intense emotions, being called "dramatic" or "emotional"

Hendrix, 2015; Leedham et al., 2020; Nichols, 2009; Wilson et. Al, 2016

- Issues with gender and not connecting with traditional gender roles

What can we do?

- Refer for further evaluation even when screening tools are subthreshold
- Compare their behaviors to those of typically developing females, NOT atypical males
- Discuss lived experiences- what are social interactions like? Are their friendships at the level of depth we would expect for their age? Are there genuine skills deficits underneath discomfort?
- Evaluate whether emotionality is tied to sensory concerns

Leedham et al., 2020; Nichols, 2009; Wilson et. Al, 2016, Sedgewick et al., 2016

Racial/Ethnic Disparities

- Autism prevalence is reported to be highest among non-Hispanic white children, lower in Hispanic, black, and Native American children, and highly variable in Asian/Pacific Islanders.
- Research hypothesizes several rea

  - Language barriers
  - Limited economic resources
  - Access to health services
  - Schedule flexibility to obtain comprehensive evaluations
  - Limited awareness of ASD and ability to report symptoms

Becerra et al., 2014; Imm, White, & Durkin, 2019; Madell, et al., 2009

Psychosocial Factors

- More intellectual disability and language delays observed in Hispanic and black children with ASD
- Black children 5.1 times more likely to be misdiagnosed with conduct disorders
- Black parents reported fewer concerns about behaviors like delayed speech and repetitive behaviors, and reported more disruptive behaviors
- Native American populations have the lowest rates of diagnosis
  - Effects of environmental racism and intergenerational trauma impacting child development

Becerra et al., 2014; Imm, White, & Durkin, 2019; Mandell, et al., 2009; Sochet et al., 2020

What can we do?

- Screen for autism in children from all backgrounds, even when the concern presented is “behavior problems”
- Increase awareness of child developmental expectations and autistic characteristics in vulnerable populations
- Education and training programs should include intentional training opportunities for culturally responsive and context specific practice, including self-examination of biases
- Education and training programs should prioritize supporting diverse students and increase representation of racial/ethnic minority clinicians in practice and leadership roles

Becerra et al., 2014; Imm, White, & Durkin, 2019; Madell, et al., 2009

Why high prevalence of gender diversity?

- Autistic people may be less influenced by social norms and so may present their internal selves more authentically
- Biological factors
- Autistic people may decide their gender identity or sexuality differently than neurotypical people

Roselli, 2018; Strang et al., 2018

What can we do?

- Respect and affirm identity and sexuality
- Those working with gender diverse populations, such as specialized clinics for gender affirming medical practices, should also screen for autism
- Clinicians working with autistic individuals should ask questions about their gender and sexual identity
- Research has shown that LGBTQA+ adolescents who have more inclusive sex education in school have better mental health
- Research and advocacy efforts should be led by the autistic community to determine how to best support these individuals

Strang et al., 2028; Warrier et al., 2020

Differential Diagnosis

ASD and Attention Deficit Hyperactivity Disorder

- Over half of individuals diagnosed with Autism also meet criteria for ADHD
- ADHD characterized by
  - Inattention: distractibility, avoiding tasks requiring sustained effort, losing things, making careless mistakes
  - Hyperactivity: fidgeting, talking excessively, interrupting others, leaving seat at inappropriate times
- Both ASD and ADHD include: problems with executive functioning (planning, organizing, completing tasks); regulating emotions; sensory processing concerns; periods of hyperfocus; social difficulties

Rommelese et al., 2010; Autism Speaks, 2017

ADHD Specific Behaviors

- Social difficulties due to not attending to social cues or excessive talking about own interests, but CAN read social cues when attending
- Does not engage in repetitive hand or body movements
- Lack of impulse control leading to intense emotionality or disruptive behaviors
- Trouble sitting still during quiet activities
- Has periods of hyperfocus but does not perseverate on interests
- Board easily and may lose interest with repetition

Cognitive and Learning Abilities

- Giftedness
  - Struggle relating to peers due to significantly higher cognitive abilities
  - Interests in topics not typical for age group
  - Asynchronous development (very advanced in some domains but behind peers in others)
- Nonverbal Learning Disorder
  - Characterized by poor visual, spatial, and organizational skills; difficulty with nonverbal cues; and poor motor performance
  - Early language skills but poor reading comprehension
  - Not an official DSM-5 diagnosis, but research support

National Association for Gifted Children; Margolis et al., 2020

Social Anxiety

- Fear of social situations or performing- worry about judgment from others and doing something embarrassing
- Avoidance of social interactions, crowded places, being around people
- Studies have estimated up to 50% of individuals with ASD also have social anxiety
- Parsing apart:
  - Developmental history- did social anxiety occur due to problems with social communication?

Broit et al., 2020

Obsessive-Compulsive Disorder and other Tic Disorders

- OCD includes
  - intrusive thoughts (sometimes specific preoccupations like fear of germs, numbers, symmetry) and
  - compulsive behaviors to manage the obsessions (excessive cleaning and organizing, counting, reassurance seeking, etc)
- Tic disorders include sudden, uncontrollable bodily movements or compulsive behaviors (vocal noises, body movements, or hair pulling)
- Prevalence of OCD in ASD ranges from 2.7 to 30%; Prevalence of ASD and Tic disorders is 10-25%
  - All may include insistence on routines or ritualistic behavior, body movements, fixations

Posteriano et al., 2017

Disordered Eating

- Some estimate 20 percent of people with anorexia are autistic
- Starvation can cause brain changes that result in autism-like behaviors, such as **social difficulties** and problems with **emotion processing**
- Difficulties with **identifying emotions** and understanding physical sensations, such as hunger, may also contribute to the overlap; common in both **autism** and **anorexia**
- For some autistic people, eating disorders may start as intense interest in calorie-counting, exercise or an insistence on a limited diet

Westwood et al., 2017

Disordered Eating Continued

- Avoidant Restrictive Food Intake Disorder (ARFID) added to DSM-5 in 2013- diagnosis reflects extreme restrictive eating but not tied to concerns of weight or body appearance
- Studies show that some autistic individuals used a dieting or attaining thinness to fit in with peer groups (as a masking or camouflaging behavior)
- Sensory concerns prompt avoidance of some foods based on textures, temperatures, etc. This can then lead to generalization of that avoidance across broad groups of foods

Lucarelli et al., 2017

Trauma

- Posttraumatic Stress Disorder
  - Characterized by intrusive thoughts about traumatic event, negative emotions and thoughts, avoidance, hyperarousal, and dissociation
  - Overlap with ASD: hyperarousal can look like sensory sensitivities; fixation on traumatic event and look repetitive; emotionality
  - Autistic individuals are vulnerable to victimization and often experience traumas – but rates of PTSD are low in this population compared to neurotypicals
- Reactive Attachment Disorder
  - Struggle to bond with caregivers; trouble managing emotion; don't seek comfort
  - Also can show poor boundaries and inappropriate behaviors towards others

Davidson et al., 2015; Gravitz, 2018

Psychosis

- Some studies show ASD increases risk of psychotic disorders (both schizoprhenia and bipolar spectrum) up to 20%
- Psychotic disorders often have later onset (late adolescence/early adulthood)
- Features of ASD are misdiagnosed as psychotic symptoms
  - Misreading others' intentions resembles paranoia
  - Problems with expressive communication resemble thought disorder
  - 'Melt downs' resemble catatonia or manic episodes
  - Both may include eccentricities in language, appearance, mannerisms; inappropriate affect

Larson et al., 2016

Borderline Personality Disorder

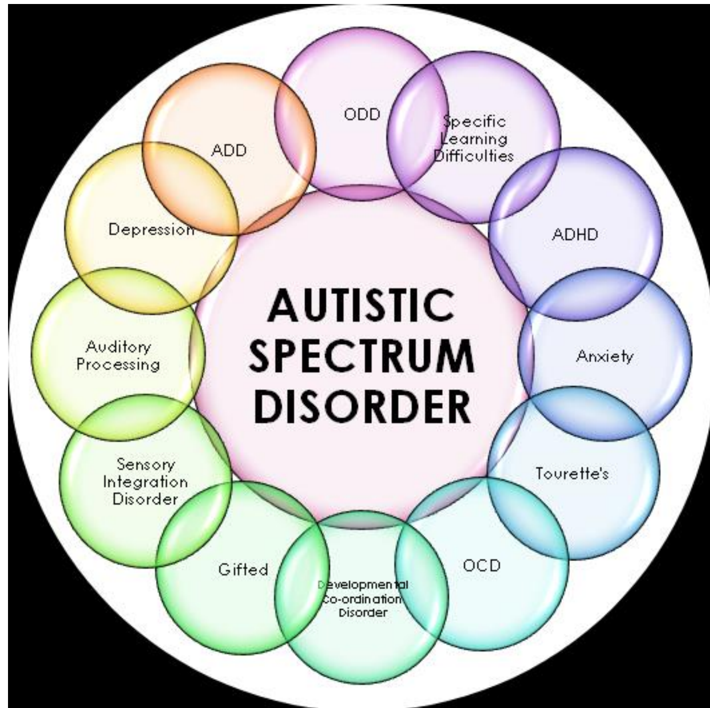
- BPD characterized by instability in interpersonal relationships, identity confusion, problems regulating emotions, impulsive and risky behaviors, suicidality
- BPD most often diagnosed in females; many women later diagnosed with ASD are misdiagnosed with BPD
- Co-morbid ASD and BPD is a group at risk for suicide, lower occurrence of substance abuse, but a more pronounced negative self image
- Some BPD characteristics look like ASD, including
  - Rigid thinking patterns (black and white thinking)
  - Emotional "meldowns"
  - Difficulties understanding other peoples' perspectives

Dudas et al., 2017

Personality Disorders

- Avoidant Personality Disorder
  - Chronic and pervasive social anxiety; extreme social inhibition; avoidance of new activities
- Obsessive Compulsive Personality Disorder
  - Perfectionistic; attention to detail that's inefficient; ridged adherence to rules/regulations; strict moral code; excessive work
- Schizoid Personality Disorder
  - Little or no desire for relationships; difficulty expressing emotions; lack of positive emotional experiences; low motivation

Anckarsäter, 2016



Remember

- Autism Spectrum Disorder encompasses many unique presentations
- ASD is better recognized overall, but still missed or delayed in vulnerable groups
- Traits of ASD overlap strongly with ADHD, Anxiety, and other psychiatric disorders
- Over 70% of autistic individuals have a co-occurring psychiatric disorder
- Increasing awareness supports early identification and linking individuals and families to much needed services
- More work needs to be done!

Autism Q&A: You asked, we answered!

Laura Rowley, PhD

Utah Center for Evidence Based Treatment

Do vaccines cause Autism

- No
- CDC highlights several research examples https://www.cdc.gov/vaccinesafety/concerns/autism.html
- Meta-analysis looking at several studies: https://www.sciencedirect.com/science/article/pii/S0264410X14006367
- Article describing the initial case study that was later refuted: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3136032/
- What to say to parents when they express this concern:

Can Autism be diagnosed in adulthood

- Yes
- BUT characteristics must be present in childhood
- Late diagnosis often occurs because characteristics may be present, but not severe, early on
- Assessments in adulthood should include corroborating data from childhood: parent/caregiver interview, school records, medical records, etc.

How do we discuss An ASD diagnosis with Adults who do not initially have that concern?

- Sometimes individuals present for other types of referral questions (like ADHD or social anxiety) and providers may suspect Autism
- In situations when the diagnosis may be unexpected, suggesting it may be challenging
- Stick to observed behaviors- what concerns are the specifically sharing that we can tie directly to signs of ASD?
- Emphasize openness- Present it as something to rule-out just in case, but that we cannot know for sure without further testing
- Connect to resources- Encourage exploration of information and see

Who diagnoses Autism?

- Psychologists or Neuropsychologists
- Developmental Pediatricians
- Pediatric Neurologists
- Child Psychiatrists
- Autism assessment requires specialized education and training!
- Asperger's/Autism Network Directory of autism diagnosticians: https://www.aane.org/directory/
- Utah Autism Evaluations: https://health.utah.gov/cshcn/pdf/Autis

How Is Autism diagnosed?

Detailed developmental history and description of concerns

- Structured diagnostic interviews
- Corroborating interviews with educators and healthcare providers

Observational measures

- IQ testing; maybe Executive Functioning testing
- Social communication and reciprocity (Autism Diagnostic and Observational Schedule, 2nd edition: ADOS-2)

Questionnaires

Why are rates increasing? Are we overdiagnosing?

- Autism diagnostic criteria did not include mild presentations (those with average to above average IQs and language abilities) until 1994 and all categories were subsumed into Autism Spectrum Disorder in 2013
- Special Education classifications did not include Autism as a separate category until 1991
- American Academy of Pediatrics recommended Autism screening at 18 month and 24 month visits in 2006

Wright, 2017

- We are more aware and increasing access to screenings for vulnerable

Why are rates in Utah so high?

- Changes in classification from previous data collection
- Improved access to resources for diagnosis
- High occurrence of risk factors associated with Autism
  - Exposure to environmental pollution
  - Pregnancies spaced less than one year apart; pregnancies in older individuals
  - Genetics

How can we improve identifying girls on the spectrum?

- Increased awareness that ASD presents differently across gender spectrum
  - Girls show less repetitive behaviors, have stronger verbal skills, more social motivation
  - Girls may not show deficits until middle school/high school when relationships are more complicated
  - Girls are better at “masking” autistic traits and “camouflaging” with peers- at great cost
- Educators and medical professionals referring for assessment, even when screening tools are below threshold

Can Autism be cured?

- There is no cure for Autism…
- …Because Autism is not a "disease," it is a broad category of neurodivergence
- However, with intervention, individuals can exhibit increased social skills and decreased repetitive/restrictive behaviors
- Diagnoses are made based on impairment…if individuals can learn to successfully navigate their world to get their needs met, there is no impairment
- Some in the ASD community argue that others are better at "masking" but it takes a LOT of energy

Where should intervention begin?

- Assuming we have a diagnosis- it depends!
- If the individual is in school- obtain educational supports
- What in daily life is hard? Focus on those areas- speech and language or occupational therapy
- Social skills groups or activities that provide opportunities for interaction
- Late diagnosis often occurs with mental health concerns- anxiety, mood disorders, etc. Psychotherapy and psychiatric medication may be important
- Autism Speaks provides guides for next steps for children and adults just diagnosed with Autism: https://www.autismspeaks.org/tool-kit

How can parents advocate for educational testing and accommodations through the school for children with ASD?

- Know your rights!
- Get advocacy support
  - States have an advocacy agency- Utah Parent Center https://utahparentcenter.org/
  - Find our local agency at Autism Society https://source.autism-society.org/autismsource/
- Get a medical diagnosis
  - Educational evaluations are not the same as a psychological evaluation that provides a medical diagnosis of Autism Spectrum Disorder

Why do individuals with ASD present with high anxiety symptoms?

- Up to 42% of children on the autism spectrum have some type of anxiety disorder
- Being neurodivergent in a neurotypical world is stressful
- Negative experiences lead to fears of those experiences (such as social interactions, navigating new situations)
- Autistic individuals prefer sameness and predictability and may react strongly to new situations, changes in plans and routines
- Autistic individuals have sensory sensitivities and may be easily overwhelmed compared to neurotypicals

In addition to sensory processing, what are other health concerns present in Autism?

- Gastrointestinal problems
  - Sensitive systems, difficulties communicating distress, sensory concerns leading to restrictive diets
- Epilepsy
  - Epilepsy affects a fifth to a third (20 to 33 percent) of people who have autism, compared to an estimated 1 to 2 percent of the general population.
- Sleep problems
  - Over half of children with autism – and possibly as many as four in five – have one or more chronic sleep problems
  - Genetics, seizures, anxiety impact sleep; sleep disturbances lead to increased

Autism Speaks Health Report
https://www.autismspeaks.org/sites/default/files/2018-09/autism-and-health-report.pdf

What can treatment providers do to support Autistic individuals with substance use disorders?

- Autistic individuals have increased risk for developing substance use disorders compared to neurotypicals, and co-occurring ASD with substance use disorder is associated with increased mortality risk
- Autistic individuals may use substances to cope with social difficulties or try to relate to peers
- Research shows the same areas in the brain activated in addiction are also activated for "stimming" and repetitive behaviors in ASD, suggesting a neurobiological link
- Treatment recommendations include less reliance on group intervention (like AA or NA) and more individualized care

Are there special considerations when conducting EMDR (Eye movement desensitization and reprossessing) with individuals with ASD?

- Guidelines for EMDR with individuals with autism https://www.emdr.nl/wp-content/uploads/2019/10/GuidelinesEMDRASD.pdf
- In general- our evidence-based treatments can be provided or adapted to autistic individuals. Many struggle with mental health concerns, and increased access is needed. Providers can build their knowledge base with appropriate continuing education, consultation, and supervision, and utilize referral networks when needed.

Thank you!

For an extensive list of resources, please visit our website: https://www.ucebt.com/resources/community/autism

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