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Primary Care Approach to Diagnostic and Management Challenges in Older Teens and Young Adults with Autism/Suspected Autism Spectrum Disorder

David Camenisch, MD/MPH

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Speaker Disclosure



- I have no actual or potential conflict of interest in relation to this program/presentation.



Learning Objectives

Attendees will be better equipped to address autism in clinical practice through:

1. Review of assessment and management strategies to address autism related behavioral challenges
2. Increased familiarity with clinical features of ASD and diagnostic challenges in older adolescent and young adult populations.



Epidemiology

- Prevalence - 1 in 68 or 1.5% across 11 communities in the United States (2012)
- Male: Female = 4-5:1
- Increased incidence likely due to a number of factors:
 - Earlier/better diagnostics
 - Increased awareness
 - Mandatory availability of treatments
 - Broader Spectrum
 - Increased understanding of association with intellectual disability and genetic syndromes (Fragile X, Down Syndrome, Fragile X, Prader-Willi, Angelman, neurofibromatosis)
 - Increased understanding of “high functioning” phenotypes
 - ... and possibly increased incidence?



Etiology

- Likely some combination of genetic susceptibility and environmental exposure \approx epigenetics
 - Specific mechanisms: methylation, acetylation, chromatin remodeling, RNA changes
- Genetic contribution – multiple mechanisms
 - Syndromic ASD (25%) - Fragile X, Prader-Willi, Angelman, Rhetts, Down, neurofibromatosis
 - Copy Number Variants (10%) – can be hereditary or idiopathic; includes duplications, deletions, inversions, translocations; > 100 identified
- Heritability – higher concordance risks for twins (identical > fraternal) and recurrence for sibling (5 – 25%) contingent on etiology
- No causal association with vaccines



Environmental Risk Factors

- Advanced maternal and paternal age
- Maternal metabolic conditions (DM, obesity, HTN)
- In utero exposures
 - Maternal infections
 - Valproate exposure
 - Traffic-related air pollution
 - Pesticides
 - SSRIs ? – recent warning to avoid in early pregnancy
- Perinatal exposures
 - Pre-term delivery
 - Low birth weight

Case #1



17-year-old male diagnosed with autism without language impairment. You are meeting patient for the first time today. There is no information regarding cognitive functioning. There has been an increase in aggression, elopement from school and suicidal ideation over the last six months starting at the end of last school year. There was some improvement over the summer but acutely worse over the last 1-2 months. He has not been sleeping well. No obvious stress at school or home. You attempt to elicit information from your patient, but he is unable or unwilling to engage. He does not currently have any MH or ASD related services. Attempts by other providers in your practice to connect them to supports have not been successful. It is not clear to you whether he would qualify for DDA services.



Where do you start?

- Set expectations for visit - acknowledge complexity of situation to parent(s), patient,...yourself.
- Give yourself permission to address what part of this you have time/information for.
- Allow yourself to not get through everything. May need multiple visits/regular follow-up.
- **How do you effectively emphasize need for mental health services and other supports?**
- **What is the differential diagnosis of behavior change in ASD?**



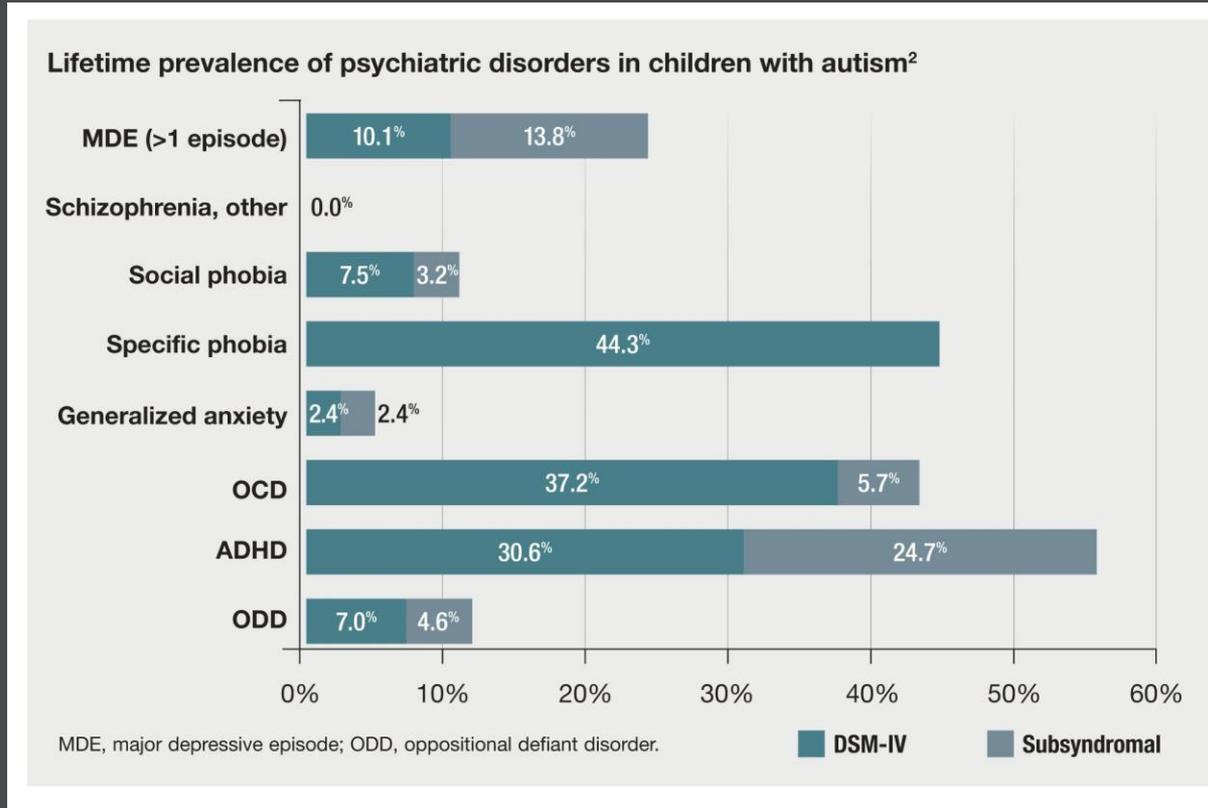
Differential Diagnosis of Behavior Change in ASD?

- bullying/social stress
- maltreatment/trauma
- medical issues (m/c = pain, GI, migraines, seizures)
- depression/anxiety
- sleep disturbance
- drug use
- environmental stress – family conflict, broken video game, financial stress, moving
- school stress – change in personnel/support, normative social stressors
- medication side effects – activation from sertraline; sedation contributing to AM/daytime irritability
-don't forget *atypical* responses to *typical* stressors is BIG part of autism.



Common Co-occurring Psychiatric Conditions in ASD

(Collins et al. Psych Times. 2019)





Schizophrenia and Autism

- Symptom overlap make distinguishing the two difficult – language difficulties, poverty of speech, formal thought disorder, over-valued ideas, interpersonal deficits, etc.
- Increasingly clear that there is shared genetic risk
- ASD confers increased risk of schizophrenia spectrum disorders ~ 10% (Jrnl Clin Med. De Giorgi *et al.* 2019)
- Prodromal and subthreshold conditions complicate later diagnosis and management both ASD and psychosis
- “atypical” versus “anomalous” perceptions
- Hallucinations are common in ASD and rarely indicate schizophrenia – reports of AH/VH relate to perceptual experiences and reporting
- Often must rely on clinical judgement due to lack of tools to evaluate psychotic symptom and sub-threshold symptoms
- Both conditions are now conceptualized as “spectrum” disorders, ASD and SSD with increasing awareness of symptom overlap and frequency of co-occurrence
- IF CONCERN FOR EMERGING PSYCHOTIC DISORDER, REFER TO PSYCHIATRIST



Mental Health Co-morbidity and Risk Factors in ASD

- 79% lifetime prevalence of psychiatric condition in adults with autism (Lever *et al.* *Jrnl of Autism and Dev Disord.* 2016.)
- 5X risk of suicide
- 10X risk of schizophrenia spectrum disorder
- Increased risk of bullying, maltreatment and all forms of abuse
- Autism traits can increase exposure to risk factors – increased rates of depression and SI due to social isolation, loneliness and feelings of being a burden
- Many conditions likely underdiagnosed and misdiagnosed due to lack of awareness, diagnostic overshadowing, symptom over-lap and lack of validated screening tools



Case #1 (continued)

Sertraline was started just before school started this year. The dose was increased to 50 mg two weeks ago. Patient's mother does not feel it has made anything worse but also not sure it has been helpful. He is prescribed trazodone as-needed for sleep. The dose was just increased to 100 mg. It is unclear if it has been helpful or how often it is given. He is also prescribed hydroxyzine 25 to 50 mg PRN anxiety. Patient's mother read about risperidone online and would like you to start it.



Medication Strategies

- Give yourself permission to practice within your comfort zone
- Whatever you do with medication(s), emphasize importance of combining with psycho-social interventions
- Possible medication interventions?
 - Thoughts about sertraline?
 - How would you address sleep?
 - Would you start risperidone?



Principles of Medication Management in ASD

- No medications are yet identified for core deficits of autism
- Medications treat related symptoms and co-occurring disorders
- Use a dimensional and symptom specific approach to evaluate incremental benefit and sustained improvement over time.
- Individuals with ASD/suspected ASD have increased sensitivity to medications in general
- “start low dose and go slow”
 - therapeutic effects seen at lower doses.
 - Monitor closely for adverse events and side effects
- Re-evaluate medication strategies frequently - no “set it and forget it”



Pharmacotherapy Options in ASD

- **ADHD symptoms/executive function deficits** - methylphenidate, amphetamines, atomoxetine, alpha-agonists, amantadine; (SSRIs)
- **Aggression / Agitation/ Irritability** - alpha agonists, AAPs, TAPs, VPA
- **Anxiety** - SSRIs, hydroxyzine, benzodiazepines, buspirone, quetiapine
- **Sleep** - melatonin, anti-histamines, alpha agonists, trazodone
- **Mood Instability** - AAPs, valproate, lamotrigine and lithium
- **Self-injury** - risperidone, naltrexone

Case #2



19 yr old female returning home from freshman year at college for holiday break. History of ADHD and social anxiety. ADHD diagnosed at 15 years of age treated with stimulant and had an IEP during high school. Not currently taking a stimulant. Socially awkward but did have a “best friend” throughout high school. Tends to gravitate towards teachers and focus intensely on some aspect of the class material. Can be emotionally reactive and tends to have difficulty maintaining relationships with female peers. Medical history significant for intermittent sleep problems, gastrointestinal issues. Patient wants help with anxiety and panic symptoms which have increased over the last few months. Mom is wondering whether her daughter has autism.



Possible directions?

- Important to distinguish between parent, patient and provider goals for encounter.
 - Focus on treatment of anxiety?
 - Re-evaluate evaluate executive function deficits?
 - Recommendations for support at school? (mental health, academic, social)
 - Pursue assessment for possible ASD?



ASD assessment in young adults

- More likely to be complex presentation
- Often on “cusp” or “fringe” of spectrum
- Presentation can be complicated by high or low cognitive capacity
- Often gaps in information regarding early developmental history – contributes to misdiagnosis
- Providers less likely to think of ASD in young adults
 - adults diagnosed with autism are more likely to raise possibility of ASD than providers (44% versus 4%) (Jones et al. Jnl Autism and Dev Disord. 2014)
- Females less likely to be diagnosed
- Reasonable to consider *whether* to pursue/give diagnosis
 - Autism as “discretionary diagnosis”
 - Consider feasibility of assessment
 - Identify goal of assessment/diagnosis – diagnosis may not add much
 - Person-centered care - consider “readiness” – How will diagnosis impact identity or sense of self?
 - *Disability versus Diversity Debate*



ASD Screening and Assessment

- Avenues for assessment/diagnosis
 - Medical specialists– pediatric neurologists, child and adolescent psychiatrists, developmental pediatricians ... and PCPs!
 - Other MH professionals: PhD, SLP
- Screening Tools – none specific to older teens/young adults
 - MCHAT (modified checklist for autism in toddlers): 16-48 months
 - CARS-2 (Childhood Autism Rating Scale): For > 2 yrs. old; 15-item, direct observation; 5-10 minutes.
- Diagnostic Aides
 - ADOS (Autism Diagnostic Observation Schedule): For toddlers to adults; direct observation, 30-45 minutes.
 - ADI-R (Autism Diagnostic Interview): For mental age > 2 yrs.; structured interview; 1.5 – 2.5 hours.
- Multi-disciplinary assessment strongly recommended for more complex presentations and older patients in general



DSM V Core Symptoms

Category A: Deficits in social communication and social interaction

1. Deficits in social emotional reciprocity
2. Deficits in nonverbal communication
3. Deficits in developing, maintaining and understanding relationships

Category B: Restricted, repetitive patterns of behaviors, interests and activity

1. Stereotyped or repetitive motor movements, use of objects or speech
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of behavior
3. Highly restricted, fixated interests that are abnormal in intensity of focus
4. Hyper or hypo-reactivity to sensory input or unusual interest in sensory aspects of environment



Other DSM V Diagnostic Highlights

- **Diagnostic changes**

- onset prior to three years of age was changed to *onset in the early developmental period*.
- sensory abnormalities was incorporated

- Inclusion of qualifiers

- with intellectual disabilities
- with language impairment
- associated with medical condition or genetic syndrome
- with catatonia

- Designation of symptom **severity level**

Level 3 – “very substantial support”

Level 2 – “substantial support”

Level 1 – “support”



Supports to consider for ASD/possible ASD

- Behavioral Support - Applied Behavioral Analysis, Pivotal Response Training
- Social Skills support – therapeutic and normative activities; social connectedness
- Speech and Language Therapy
- Occupational Therapy
- Mental Health Treatment
- Developmental Disabilities Administration services –requires cognitive testing and assessment of adaptive function (Vineland, ABAS)
- Transition planning – to/through adulthood; facilitate autonomy; guardianship; supported housing; assistance applying for SSDI
- Vocational Rehab/job placement assistance
- Caregiver support/Respite – consider systems of care and natural supports
- Normative/normal activities – not everything has to be explicitly therapeutic



General Management Strategies

- Distinguish between patient, caregiver and provider goals
- When possible, start with patient goals and identified needs
- Okay to defer assessment/diagnosis until more clear picture
- Treatments/interventions should target goals and skills deficits regardless of diagnosis
- Multidisciplinary care is the rule rather than the exception
- Type of support needed will likely shift over time depending on individual strengths and vulnerabilities – schedule regular check-ins
- Advocacy may be necessary to assemble a team and maintain momentum.
- It takes a team! You can't do it alone.
- It takes time! You can't do everything in one visit.
- Be patient! - “Moving the needle” clinically and accessing services can be a very slow process.



ASD Support Across the Lifespan

Washington Autism Alliance www.washingtonautismalliance.org

Arc (by County or Region) www.thearc.org/chapter/the-arc-of-washington-state/

Seattle Children's Autism Online Resource Center

www.seattlechildrens.org/clinics/autism-center/patient-family-resources/

Alyssa Burnett Center Adult Life Center

<https://www.seattlechildrens.org/clinics/autism-center/alyssa-burnett-adult-life-center/>

UW Adult Autism Center

<https://www.uwmedicine.org/locations/adult-autism>

PAL guide <https://www.seattlechildrens.org/globalassets/documents/healthcare-professionals/pal/wa/wa-autism-care-guide.pdf>



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