ECHO AUTISM COMMUNITIES

Advanced Diagnosis

1

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Project ECHO

CODA: Differential Diagnosis in Youth and Adults

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2

ECHO Autism: Advanced Diagnosis Continuing Education

Target audience: Psychologists and Physicians

Objective: Increase local access to high-quality more complex, Autism Spectrum Disorder (ASD) diagnostic evaluations. Diagnosticians will increase their self-efficacy, knowledge, and skill in autism best practices for evaluation and differential diagnosis.

Successful Completion

- Successful completion of this activity, which includes participating in the educational offering, participating in the evaluation process, and completing the verification of attendance, enables the learner to satisfy the requirements for continuing education.

Continuing Medical Education (CME)

- *The University of Missouri - Columbia School of Medicine is accredited by the Accreditation Council for Continuing Medical Education (ACCME) to provide continuing medical education for physicians.*
- The University of Missouri - Columbia School of Medicine designates this **live** educational activity for a maximum of ***1.5** AMA PRA Category 1 Credit(s)™*. Physicians should only claim the credit commensurate with the extent of their participation in the activity.

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ECHO Autism: Advanced Diagnosis Speaker Disclosure

Relevant Financial Relationship Disclosures

Current ACCME (Accreditation Council for Continuing Medical Education) rules state that participants in CE activities should be made aware of any relevant affiliation or financial interest in the previous 24 months that may affect the planning of an educational activity or a speaker's presentation(s). Each planning committee member and speaker has been requested to complete a financial relationship reporting form for the **ECHO Autism: Advanced Diagnosis Series.**

Speaker Disclosures:

Kristin Sohl, MD,FAAP receives support:

- **Cognoa Behavior Health – research support**
- **Quadrant Biosciences – medical science collaborator**

Valeria Nanclares-Nogues, PsyD has the following relationships:

- **WPS – as an independent certified ADOS-2 and ADI-R trainer**
- **TEA Ediciones – as an independent certified ADOS-2 and ADI-R trainer**
- **Vanderbilt University – as an independent certified STAT trainer**

All relevant financial relationships for the presenter(s) have been mitigated.

No other speaker or planning committee member has a relevant financial interest

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Objectives

- Review of Differential Diagnosis vs Comorbidity
  - Knowledge burst will focus on differential diagnosis, not co-occurrence
- Describe ID vs ASD
- Describe Social Anxiety vs ASD
- Describe OCD vs ASD

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5

Differential Diagnosis vs. Comorbidity

- **Differential Diagnosis:**
  - Process of determining which (of two or more) distinct disorders is present
  - ASD vs. Other Condition
- **Comorbidity:**
  - Two (or more) distinct disorders are present in the same individual
  - ASD + Other Condition
- **Symptom Overlap:**
  - Symptoms may be shared by multiple conditions
  - Some symptoms may mimic ASD symptoms

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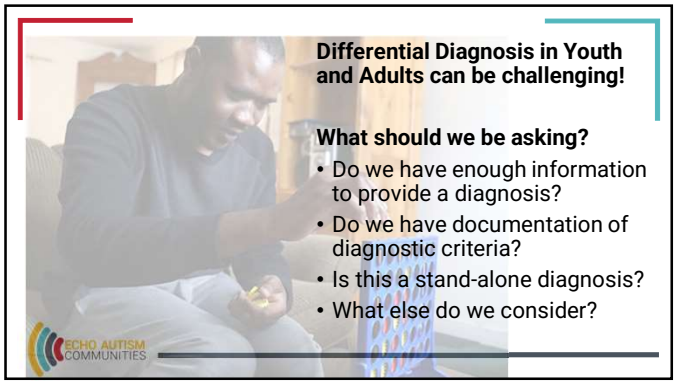
6


Differential Diagnosis in Youth and Adults can be challenging!
What should we be asking?
• Do we have enough information to provide a diagnosis?
• Do we have documentation of diagnostic criteria?
• Is this a stand-alone diagnosis?
• What else do we consider?
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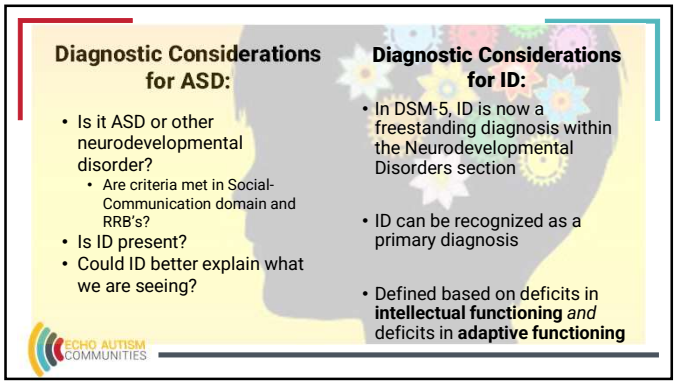
Diagnostic Considerations for ASD:
• Is it ASD or other neurodevelopmental disorder?
• Are criteria met in Social-Communication domain and RRB's?
• Is ID present?
• Could ID better explain what we are seeing?
Diagnostic Considerations for ID:
• In DSM-5, ID is now a freestanding diagnosis within the Neurodevelopmental Disorders section
• ID can be recognized as a primary diagnosis
• Defined based on deficits in intellectual functioning and deficits in adaptive functioning
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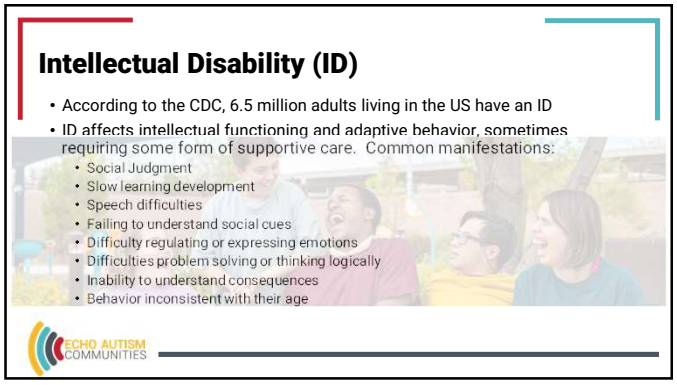
Intellectual Disability (ID)
• According to the CDC, 6.5 million adults living in the US have an ID
• ID affects intellectual functioning and adaptive behavior, sometimes requiring some form of supportive care. Common manifestations:
• Social Judgment
• Slow learning development
• Speech difficulties
• Failing to understand social cues
• Difficulty regulating or expressing emotions
• Difficulties problem solving or thinking logically
• Inability to understand consequences
• Behavior inconsistent with their age
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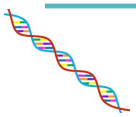
9

ASD vs ID



- One of the most difficult differential diagnoses.
- Genes associated with ASD are often the same genes associated ID (Casanova, et al., 2016; Zhu, et al., 2014)
- Considerable overlap between ASD and ID both etiologically and phenotypically (Sanders et al., 2015; Robinson, et al., 2014)
- So, what are the most distinguishing diagnostic features?

The short answer, basic social communication skills!

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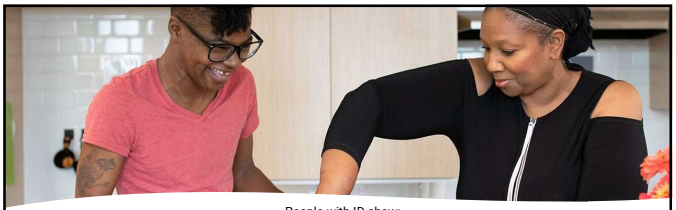
10



For ASD, social deficits exceed the expected deficits for that intellectual level.

- People with ID show:
  - Greater capacity for joint attention behaviors
  - Range of directed affect
  - Use of socially appropriate gaze
  - Immature but socially reciprocal behavior
- Autistic People show:
  - Difficulties with joint attention behaviors
  - Decreased use of gaze to regulate interaction
  - Decreased social-emotional reciprocity
  - Decreased non-verbal communication skills

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11

How to know if social communication is below the expected for his/her general developmental level

- Consider behaviors expected for a given developmental level
- Consider chronological age as well: life experiences on behaviors (e.g., sit for longer periods of time, more expertise with certain objects/devices/toys, compliance skills)

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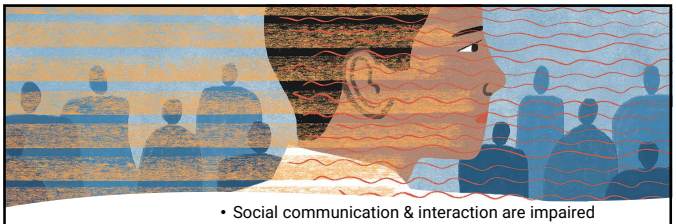
12



When do ASD and ID co-exist?

- Social communication & interaction are impaired ***relative to the developmental level*** of the individual's nonverbal skills
- When ASD and ID co-exist, **adaptive skills** are seen below what is expected based on IQ, becoming even more impaired and with less functional independence

(Ashwood et al., 2015; Pugliese et al., 2015; Kanne et al., 2011; Duncan et al., 2015).

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13

How do we distinguish ASD from Social Anxiety?

First let's ask:
What is the primary deficit?

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14

Same, but quite different!

Both experience difficulties in social situations

- Atypicalities in social attention (i.e., avoidance of gaze and social interactions)
- Social skills deficits:
  - Body posture
  - Eye contact used in interactions
  - Speech qualities
- Trouble building and maintaining friendships

Kleberg et al., 2017

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15

So, what makes them different?

Autism Spectrum Disorder

- In ASD, challenges are seen from a young age
- Impairment in social situations is due to difficulty reading and understanding social and emotional cues
- Presence of restricted and repetitive behaviors
- May prefer to be by themselves

Social Anxiety

- Symptoms generally emerge in adolescence or there is worsening of symptoms at this time.
- Driving force is anxiety in social and performance situations – limited by anxiety
- No Restrictive and repetitive behaviors are found
- Typically desire social interaction

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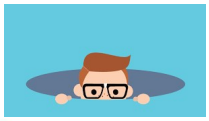
16

Social Anxiety is driven by fear

What will others think?
What will they say?
Fear of performance
Fear of exposure
Fear of ridicule



Relationships are impaired by anxiety!

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17

Final Thoughts: What do they look like?

Social Anxiety

- Fear of embarrassment or humiliation
- No difficulties understanding jokes, humor, or double meaning
- Often overly sensitive to body language of others
- Stand too far from others
- Often speak too softly

ASD

- Seen as rude – Theory of Mind!
- Don't get double meaning
- Difficulty taking hints or understanding humor
- Difficulty understanding meaning of gestures, tone of voice, facial expressions
- Personal space issues – too close
- May talk too loudly – lack of modulation

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18

Differential Diagnosis between ASD and OCD

- Similar symptoms, in terms of rigidity and repetition.
- OCD rituals can resemble the repetitive behaviors common in autism
- People with autism are twice as likely as those without to be diagnosed with OCD later in life (Meier et al., 2015)
- Overdiagnosis of OCD in ASD (Paula-Pérez, 2013)

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19

The key to understanding lies in the differences!

- ASD and OCD are both classified as neurological disorders.
- They share many similarities:
  - anxiety
  - intrusive thoughts, urges and feelings
- **But, there are some crucial differences!**

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20

ASD is driven by sameness

- Early onset social communication deficits
- ASD symptoms present earlier than OCD symptoms
- Obsessive-compulsive behavior is EGOSYNTONIC (may even be pleasurable)
- There is a desire to keep things the same
- No perceived responsibility; no attempts to neutralize; may not even be aware of doing so

Meng-Chuan Lai y Simon Baron-Cohen (2015)

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21

OCD is driven by intrusive thoughts

- Awareness that it is not rational
- Feel trapped or stuck in their worries
- They come out of nowhere
- Often cause a great deal of anxiety
  - Worries about getting hurt
  - Suspicion that partner is unfaithful

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ASD vs OCD: Main differential

- How the individual **reacts** to repetitive or compulsive actions and tendencies in public!
- An individual with OCD will be ashamed of his or her actions.
- An individual on the spectrum is not usually affected by what others think.
- In ASD there are social impairments, not so in OCD

(Harris, 2018; Stairway to STEM)

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23

So, what makes them different?

Autism Spectrum Disorder

- Characterized by need for sameness, difficulty with change
- Presence of restricted and repetitive behaviors as hallmark
- ASD individuals tend to struggle in social situations

OCD

- Characterized by persistent, distressing thoughts.
- Engages in compulsive behaviors to manage those thoughts
- Consistent repetitive actions (e.g., need to touch objects repeatedly, need for certain sequences of behavior, etc.)
- Typically do not struggle in social situations

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24

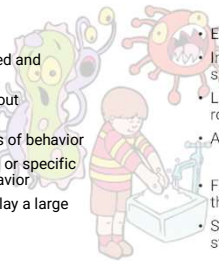
Final Thoughts: Distinction is easier said than done

ASD

- EGOSINTONIC
- Repertoire of restricted and repetitive behaviors
- Anxiety may bring about repetitive behaviors
- May not be conscious of behavior
- No intrusive thoughts or specific worries drive the behavior
- Sensory challenges play a large role

OCD

- EGODISTONIC
- Intrusive thoughts lead to specific behaviors
- Lack of performing specific routines causes anxiety
- Awareness it is not rational
- Feels trapped or stuck in their worries
- Sensory issues may be the start of a worry/concern



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25

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- https://www.stairwaytostem.org/how-to-tell-if-you-have-autism-ocd-or-both/

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26

Genetic Syndromes/Disorders

https://www.autism.org/related-disorders/

- Similar Behavioral Features and/or Common Comorbidity with ASD:
  - Fragile X Syndrome – Major genetic cause of autism
    - Those w/o share some features – poor EC, difficulties in social situations (although often worse in ASD), repetitive behaviors (e.g., hand flapping, but not broader scope of RRBs), ID https://www.spectrumnews.org/news/fragile-x-syndromes-link-autism-explained/
  - Tuberous Sclerosis – 40-50% have ASD - Major genetic cause of autism
    - Other neuropsychiatric disorders associated with TSC: ID and Behavioral difficulties. 85% have seizures
    - Epilepsy, brain tumors, and kidney tumors are more life-threatening manifestations
    - https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4668631/
  - Landau-Kleffner Syndrome – acquired epileptiform aphasia
    - Normal language and cognitive development before the onset of sx and normal hearing
    - The younger the child at age of onset, the worse the prognosis
    - Shared presentation with regressive form of ASD
    - https://austinpublishinggroup.com/autism/fulltext/autism-v2-id1031.php
    - Rett Syndrome –Mutations of MECP2 gene on X chromosome – virtually all girls
    - Repetitive hand movements, loss of language and motor skills (even walking), withdrawal – in ASD RRBs more varied, with cognitive involvement as well – rituals, routines, intense focus/interest
    - A period of apparently typical development, followed by loss of skills (bw 6-18 months)
    - https://www.spectrumnews.org/news/rett-syndromes-link-to-autism-explained/
  - Cornelia de Lange Syndrome – Rare genetic disorder often accompanied by ASD – DD, abnormal facial/physical features, short stature, epilepsy, and ASD
    - Social differences less pronounced than ASD
    - Less likely to have pronounced RRBs, more likely to use EC and gestures to communicate, more anxious
    - https://www.spectrumnews.org/news/cognition-and-behavior-rare-syndrome-distinct-from-autism/

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27

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28

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29