AUTISM SPECTRUM DISORDERS: DIAGNOSIS AND MEDICAL EVALUATION

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ASD AND DSM-V – HOW IS IT DIFFERENT?

- No longer diagnostic subcategories – Autism, PDD NOS and Aspergers
- Rett's and CDD are no longer part of ASD
- Behavioral criteria can now be met by historical report as well as direct observation.
- All 3 Social Communication and Social Interaction criteria must be met PLUS at least 2 of the Restrictive or Repetitive Behaviors/Activities/Interests criteria
- Social (Pragmatic) Communication Disorder should be considered as an alternative for patients who have social communication/interaction deficits, but don't meet other ASD criteria.
- Symptoms do not have to be present before age 3!
- Must cause clinically significant impairment (Level 1, 2, 3)
- Not better explained by GDD or intellectual disability

SOCIAL COMMUNICATION AND SOCIAL INTERACTION CRITERIA

- All 3 criterion must be met for diagnosis:
- 1. Deficits in Social-Emotional Reciprocity
- 2. Deficits in Non-verbal Communicative Behaviors used in social interaction
- 3. Deficits in Developing, Maintaining and Understanding social relationships

RESTRICTED, REPETITIVE BEHAVIOR/INTERESTS/ACTIVITIES CRITERIA

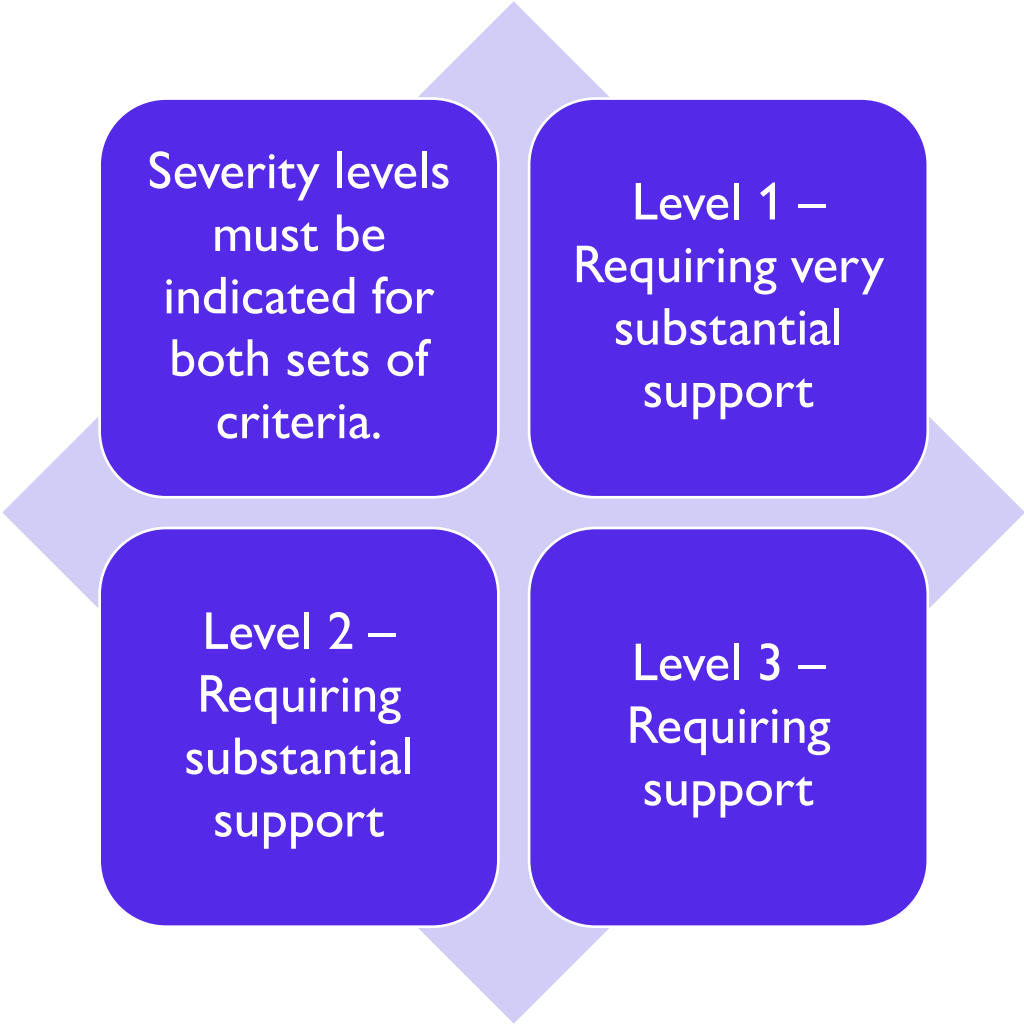
Stereotyped or repetitive motor movements, use of objects or speech

Insistence on sameness, inflexible adherence to routines, or ritualized patters of verbal and/or non-verbal behaviors

Highly restricted, fixated interests that are abnormal in intensity or focus

Hyper- or hypo-sensitivity to sensory input or unusual interests in sensory aspects of the environment

LEVELS OF SUPPORT



HOW IS IDEA DIFFERENT

- "Autism means a developmental disability significantly affecting verbal and nonverbal communication and social interaction, generally evident before age three, that adversely affects a child's educational performance."
- Doesn't use term ASD
- Does not apply if symptoms in educational environment are due to ED, per IDEA definition
- Does not apply if symptoms don't impact the child's ability to succeed in an educational environment.

SOCIAL COMMUNICATION DISORDER

- Persistent deficits in verbal and non-verbal communication in ALL 4 of these categories:
  - Deficits in using communication for social purposes in a manner appropriate for social context
  - Impairment in ability to change communication to match context or listener needs
  - Difficulty following rules for conversation and storytelling
  - Difficulty understanding what is not explicitly stated and nonliteral or ambiguous meaning of language
  - Deficits impact functioning, onset in early childhood, and not explained better by another neurologic diagnosis (ASD, GDD, ID)

COMPONENTS OF EVALUATION

- Structured History - developmental, medical, sleep/eating, behavioral, family history
- Direct Observation – free play, structured play, engagement with parent
- ASD-Specific Diagnostic Tools
- General Physical and Neurologic Exam – head circumference, skin exam
- Medical Testing, as indicated
- Other very helpful data:
    - Information from the classroom environment (BASC)
    - Psychoeducational testing – cognitive assessment, achievement testing
    - Adaptive Behavior scales (Vineland)
    - Speech and Language evaluation – including assessment of Pragmatic language

ASD DIAGNOSTIC TOOLS

- Autism Diagnostic Observation Scale (ADOS-2) – training recommended; can be integrated into a clinic visit; considered a "gold standard"
- Autism Diagnostic Interview (ADI-R) - really, really long; considered a "gold standard"
- Childhood Autism Rating Scale (CARS-2) – clinician scores child during observation
- Gilliam Autism Rating Scale (GARS-3) – has parent and teacher scales
- Social Responsiveness Scale (SRS) – impact of symptoms in natural social settings

MEDICAL TESTING FOR EVERY CHILD WITH ASD

- Audiology evaluation
- Microarray
- Fragile X testing

*Genetic testing yields about a 20% positive rate, though can be more in individuals with GDD, Intellectual Disability, seizures or dysmorphology.

TARGETED MEDICAL TESTING TO CONSIDER

- Metabolic testing – vomiting, FTT, hypotonia, lethargy, regression, multiple organ-system involvement or movement disorder (SAA, UOA, lactate/pyruvate, carnitine, acylcarnitine profile and CMP). Lysosomal testing if organomegaly present.
- Lead and ferritin levels – pica; at risk home environments
- EEG – acute developmental regression, staring spells, unexplained behavior change
- MRI – acute regression, abnormal neuro exam, midline facial changes, microcephaly, neurocutaneous stigmata
- Additional genetic testing
- Testing recommended for co-morbid conditions – i.e. sleep study

GENETIC DISORDERS ASSOCIATED WITH ASD

- Angelmans –seizures, happy affect, hand waving or clapping, intellectual disability, LE spasticity; 15q AS/PWS site (note: microarray may be normal!)
- Bannayan-Riley-Ruvalcaba – macrocephaly, lipoma, penile freckling, macrosomia – PTEN gene; Increased risk of breast, colon, thyroid cancers
- Trisomy 21/Down Syndrome – up to 1 in 10 may have ASD behavioral profile
- Fragile X – macrocephaly, long facies, low-set ears, joint hypermobility, testicular enlargement (post-puberty); FMR1 gene; pre-mutation risks
- Mitochondrial disorders – hypotonia, FTT or poor growth/feeding problems, regression, seizures
- Rett Syndrome – normal development then regression, seizures, hand automatisms, progressive microcephaly; MECP2 gene
- Smith Lemli Opitz – intellectual disability, syndactyly or webbing, broad flat nasal bridge, may have multi-organ involvement; 7-dehyrocholesterol is elevated in serum

AND DON'T FORGET…

- Plan for behavioral intervention – ABA, IEP planning, Psychology services
- ST/OT referrals, as indicated
- Discuss evaluation or treatment plan for other co-morbidities – sleep, eating, toileting, ADHD, anxiety, mood disorders, movement disorders, seizures, etc. **More on this topic next time!**
- Parent supports – ASA, Autism Speaks, local clubs (i.e. Orchid club, Bozeman)

ANY QUESTIONS?

Feel free to reach out to me directly!

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