

CO-OCCURRING DOWN SYNDROME AND AUTISM SPECTRUM DISORDER: RISK FACTORS, RESEARCH, AND RESOURCES

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Objectives

- Define Down syndrome (DS) and autism spectrum disorder (ASD)
- Overview of co-occurrence (DS-ASD)
- Risk factors/symptoms
- Evaluation for autism spectrum disorder
- Recommendations and Resources

Definitions

Down Syndrome

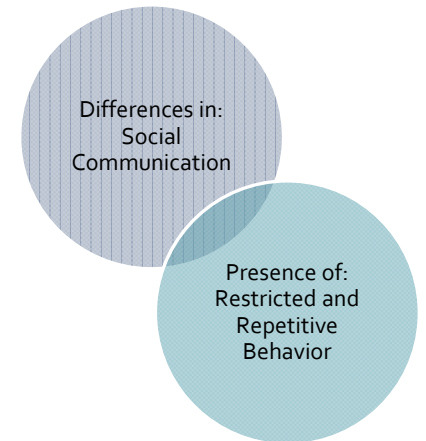
- Genetic/medical diagnosis
- Caused by presence of full or partial trisomy of chromosome 21
- Intellectual disability mild-moderate range
- Repetitive behaviors common

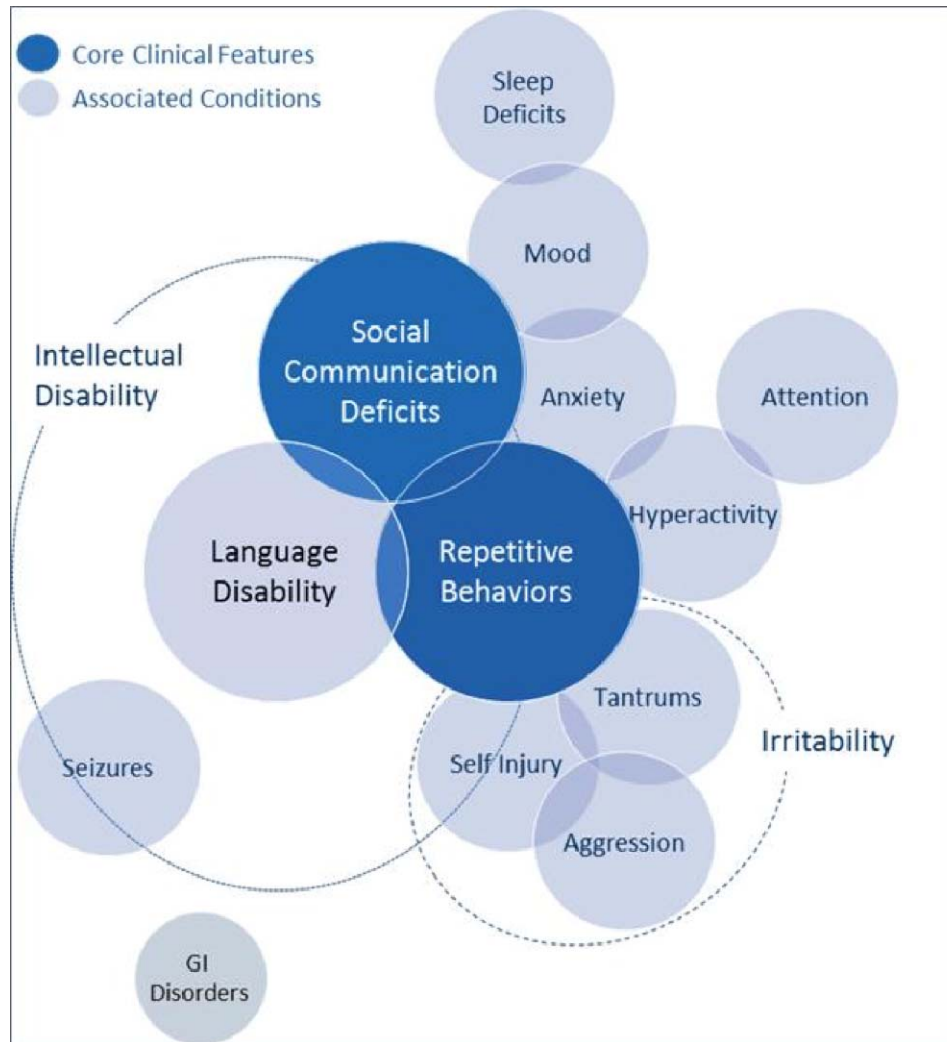
Autism Spectrum Disorder

- Behavioral diagnosis
- Multi-factorial cause
- Characterized by differences in social communication and the presence of restricted and repetitive behaviors
- No biological test available

So what is autism, really?

- Based on the DSM-5¹
 - Persistent deficits in social communication and social interaction across environments
 - Restricted, repetitive patterns of behavior, interests, or activities
 - Symptoms present early in development
 - Symptoms cause impairment across environments
 - *Symptoms not better explained by intellectual disability or global developmental delay*





Klinger, L., Dawson, G., Burner, K., & Crisler, M. (2014)

Facts and figures for ASD

1 in 59 children

4 times more
common in males

1 in 10 also have
genetic condition
(DS, FXS)

2 years of age
identification most
reliable

4 years of age
average age of
identification

150% increase in
prevalence
between 2000 and
2014

What does DS-ASD look like?

- Co-occurrence of ASD and DS ranges from 5% to 39 %²⁻⁵
- May have greater intellectual impairment than DS alone ^{2, 6-7}
 - ASD symptoms are above what is explained by intellectual impairment
- Higher rates of stereotyped behavior, repetitive use of language, over-activity ^{2, 6-7}
- Poor social orienting, infrequent social overtures, limited shared affect ⁶
- Differences in functional play⁶
- Research limited in this area across the lifespan

Risk Factors for DS-ASD

- Infantile spasms
- Complications of heart surgery
- Early hypothyroidism
- Male sex
- Regression reported in up to 50% of individuals ⁸
 - Later than regression observed in ASD (can be as late as 5 years) ⁹
 - Leads to delays/difficulty with identification of ASD
 - Supports need for ongoing screening after age 2

When to ask the question?

- Ask whenever you are concerned or want to know!
- Diagnostic overshadowing- when one diagnosis interferes with the detection of the other diagnosis because of the generalization that “those symptoms are just due to Down syndrome”
- Focus on the *absence* of behaviors rather than the presence
 - For example, lack of initiating social interactions more concerning than presence of hand flapping

What does an evaluation for ASD involve?

- Medical evaluation
- Cognitive development/IQ
 - Social communication function must be qualitatively different than general cognitive function
- Adaptive/daily living skills
- Language abilities
 - Receptive, expressive, pragmatic
- Use of standardized tools to assess ASD symptoms
- Consider developmental course and differential diagnosis
 - features of withdrawal that emerge in adolescence may have other cause

Why does it matter?

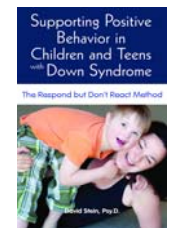
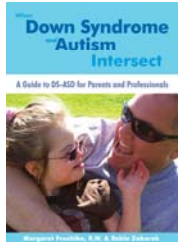
- Many developmental issues similar, other areas such as social development more impaired in DS-ASD
- Education may look different with increased focus on social skills
- Additional medical work-up may be recommended with DS-ASD
- Can affect IFSP/IEP classification and related services
 - Recommend primary eligibility recognize ASD
- Increased social support from other families

What's next?

- Need for both family support and child support
 - Some families will choose to initiate Applied Behavior Analysis or ABA therapy for autism
 - Intensive level of services for younger children (comprehensive)
 - Less intensive for school-aged children (focused)
 - Communicate findings with school team and other treatment providers
 - Communicate with primary care physician

Resources

- *“When Down Syndrome and Autism Intersect: A Guide to DS-ASD for Parents and Professionals”* by Margaret Froehlke, R.N., & Robin Zaborek
- *“Supporting Positive Behavior in Children and Teens with Down Syndrome: The Respond but Don’t React Method”* by David Stein, Psy.D.
- Wisconsin Regional Centers Children and Youth with Special Health Care Needs (CYSHCN) <https://www.dhs.wisconsin.gov/cyshcn/regionalcenters.htm>



Resources



WAISMAN
CENTER

- Waisman Center Clinics 608-263-3301
- “Dynamic Duals” family group sponsored by MADSS and GiGi’s Playhouse
- Autism Internet Modules
 - Provides professional development on strategies for treating symptoms of ASD
 - www.autisminternetmodules.org
- Autism Distance Education Parent Training (ADEPT) Modules- UC David
 - Online learning for parents to teach children with ASD and other DD
 - http://ucdmc.ucdavis.edu/mindinstitute/centers/cedd/cedd_adept.html
- Autism Focused Intervention Resources and Modules (AFIRM)
 - <https://afirm.fpg.unc.edu/>

References

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- ²Capone, G.T., Grados, M.A., Kaufmann, W.E., Bernad-Ripoll, S., Jewell, A. (2005). Down syndrome and co-morbid autism spectrum disorder: Characterization using the aberrant behavior checklist. *American Journal of Medical Genetics* 134, 373-380.
- ³DiGuiseppi, C., Hepburn, S., Davis, J.M., Fidler, D.J., Hartway, S...et al. (2010). Screening for autism spectrum disorders in children with down syndrome population prevalence and screening tests characteristics. *Journal of Developmental and Behavioral Pediatrics*, 31, 181-191.
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- ⁵Moss, J., Richards, C., Nelson, L., & Oliver, C. (2012). Prevalence of autism spectrum disorder symptomatology and related behavioral characteristics in individuals with Down syndrome. *Autism*, 17(4), 390-404.
- ⁶Carter, J.C., Capone, G.T., Gray, R.M., Cox, C.S., & Kaufmann, W.E. (2007). Autistic-spectrum disorders in down syndrome: Further delineation and distinction from other behavioral abnormalities. *American Journal of Medical Genetics Part B*, 114B, 87-94.
- ⁷Molloy, C.A., Murray, D.S., Kinsman, A., Castillo, H., Mitchell, T...et al., (2009). Differences in the clinical presentation of Trisomy 21 with and without autism. *Journal of Intellectual Disability Research*, 53, 143-151.
- ⁸Hickey, F. & Patterson, B. *Occurrence of language regression and EEG abnormalities in children with Down syndrome and autism spectrum disorders*. Paper presented at: International Meeting for Autism Research; May 5-7; Boston, MA.
- ⁹Castillo, H., Patterson, B., Hickey, F., Kinsman, A., Howard, J.M...et al. (2008). Difference in age at regression in children with autism with and without down syndrome. *Journal of Developmental and Behavioral Pediatrics*, 29, 89-93.

Questions?