DSMIG-USA®
Down Syndrome Medical Interest Group

# Evaluating Autism in Children with Down Syndrome

Down Syndrome Medical Interest Group: DS-ASD Workgroup

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Autism is common in Down syndrome (DS): approximately 16-18% of persons with DS also have autism. Caregivers may bring up concerns about their child's communication, behavior, and social skills to their providers and educators. Other times, professionals may note signs of autism that prompt further evaluation. This document aims to help professionals as they consider a possible co-occurring autism diagnosis.

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## The Medical Work-Up

It is important to correctly identify co-occurring diagnoses (both neurodevelopmental and medical) when they are present as this can lead to more informed treatment options, potential access to services, and improved quality of life. A thorough review of medical history and medical work up is recommended when considering a diagnosis of autism in a person with Down syndrome (DS-ASD).

Medical conditions that are common in Down syndrome can present as "autistic-like" characteristics/behaviors. Medical conditions do not exclude a possible diagnosis of autism.

Follow the American Academy of Pediatrics Guidelines for healthcare maintenance. Be aware of diagnostic overshadowing (attributing symptoms to Down syndrome rather than to a potential co-occurring condition).

*Bloodwork*

- TSH/FT4
- CBC
- Lead
- Celiac screen (can present with behavioral symptoms in children with DS)

Consider possible referral for genetic testing as a reasonable consideration for patients with DS and features suggestive of a secondary diagnosis, such as autism.

*Evaluation for conditions that can impact social communication and behavior:*

- Vision exam by an optometrist or ophthalmologist
    - Whenever possible, include assessment for cerebral visual impairment (CVI) (This requires functional vision assessment in addition to the eye exam.)
- Hearing assessment with a pediatric audiologist.
    - May require behavioral support or ABR
- Assess for sleep apnea
    - This requires a sleep study, which may take a while due to waitlists. You do not need to wait for the sleep study results to trigger an autism evaluation.
- Assess for presence of a seizure disorder (if indicated)
- If there is regression after complicated surgery, consider r/o brain injury as a complication post-surgery.

*Some medical conditions appear to occur more commonly in children with DS and autism. Evaluate frequently and treat co-occurring:*

- Constipation and reflux
- Swallowing difficulties and dysphagia

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- If profound motor delays are present, consider further evaluation that may include an orthopedic assessment.
- Sleep disturbances not due to sleep apnea (e.g., delayed sleep latency, early morning awakening)

## Behavioral Health Assessment

While many individuals with DS will have some degree of impairment in one or more of the areas of development listed below, individuals with DS-ASD are typically more impacted and in multiple areas concurrently. It is recommended providers review DSM-5 criteria for Autism Spectrum Disorder when considering referral for evaluation.

It is vital to observe and listen to the individual's caregiver, health/education team members, and your own impressions if the child is presenting differently from other children with DS. Caregivers may notice differences between their child and peers with DS related to social communication and behavioral functioning. The questions below may help guide next steps (e.g., referral for futher evaluation if indicated).

Request and review prior IEP/functional behavioral plans, therapy reports, and other records that may be available.

It can be helpful to request/review videos from the child’s caregiver of their child in different settings, particularly showing how the person interacts with others (e.g., siblings, peers, teachers).

**Gather Information related to potential indicators of autism:**

- Individual's social motivation (Do they need support or prompting to use gestures or pointing? Do they tend to keep to themselves and not initiate social interactions?

- Behavioral concerns (ask about self-injury, meltdowns, aggression)

- Sensory differences
    - Endorsement of sensory seeking behaviors/interests – preference towards certain textures, smells, tastes and movement, proprioceptive input (stimming)
    - Endorsement of sensory aversion behaviors - haircuts, toothbrushing, bloodwork; avoidance of specific textures for clothing, foods; sensitivity to noise, crowds

- Use of verbal and non-verbal language (Languages delays are not in and of themselves indicative of an autism diagnosis)
    - Spontaneous language - Do they require prompting to communicate?
    - Socially directed language - Do they use language largely to label things rather than to request or share with others?
    - Language abilities/delays - Are there profound language delays in the absence of severe hearing loss?
    - Repetitive language/sounds - Do they use phrases/words to perseverate on a sound or word rather than communicate, or repeat words?

- Do they have a special intense interest to the exclusion of other topics or toys?

- Nonverbal communication – do they avoid eye contact with speaker?

**Observe:**

- How intense are repetitive behaviors – are they difficult to redirect or interrupt?
- How severe are the sensorimotor behaviors/ stimming?
- How difficult is it to engage the child and sustain that engagement?
- How does the child initiate interactions with you or their caregiver(s)?

- How do they play with toys? Do they mostly play repetitively or do they use the toy functionally or symbolically?
- How do they vocalize and do they integrate vocalizations with gestures, eye contact?

**Screen:**
Complete further screening of social engagement. May need to consider non-standardized administration (outside of age-related norms). Potential options may include:

  - Modified Checklist for Autism in Toddlers -Revised (MCHAT-R/F)
  - Survey of Wellbeing of Young Children (SWYC)
  - Social Communication Questionnaire, 2nd Edition (SCQ-2)
  - Social Responsiveness Scale, 2nd Edition (SRS-2)

**Refer:**
Refer families for a comprehensive diagnostic evaluation for co-occurring autism. When available, refer to a local autism center of excellence, university center on disabilities, DS clinic, or hospital, while being mindful of wait times for evaluations (some of these highly specialized centers may have long waitlists). If specialized clinics are not available, refer to a psychologist in the child's insurance network. An educational assessment for autism can be requested at the same time if the child is in school. Whenever possible, an in-person evaluation is preferred over a telehealth evaluation.

- While waiting for the ASD assessment, connect the child and the family with their local county/regional birth-to-three early intervention programs, speech therapy, occupational therapy, and physical therapy. If referral to evidence-based intervention can be initiated without a formal diagnosis of ASD, consider referral while waiting for the ASD assessment.

- If not already in an educational or school program, discuss enrolling the child in a structured program through their local county/regional centers and daycare or preschool program.

## Advocate and Support (Post Diagnosis)

- A diagnosis of ASD can allow access to more school-and community-based services.
- Advise families to speak to the Early Intervention (0-3 years) or Special Education Director (3+ years) about the new diagnosis of DS-ASD and share their evaluation report to revise their treatment plan.
- Share information about ASD in DS with the family and education/health team members.
- Provide families with the information for The Down Syndrome-Autism Connection.
- Inform families of evidence-based behavioral treatments for ASD.
- When Down Syndrome and Autism Intersect: A Guide to DS-ASD for Families and Professionals (Passion Flower Press, 2/2024)
- "Autism and Down syndrome" – Down Syndrome Center Podcast (https://downsyndromecenter.libsyn.com/80-autism-and-down-syndrome-with-dr-nicole-baumer)

*** Additional resources to gain more information on Down syndrome and autism (DS+ASD) and many of the topics included in this document are available on the DSMIG website.