Autism & Down Syndrome:

How to recognize the signs and support your child

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Questions to Answer:

- What's the difference (Autism vs. Intellectual Disability)?
- How common is autism in Down syndrome?
- What does autism look like in a child with DS — and how is it different from "just DS"?
- Red flags: What should parents watch for?
- Why does a dual-diagnosis matter?
- What to do next? — practical steps

Definitions

Intellectual Disability

Challenges with both:

- Intellectual functioning
  - Reasoning
  - Problem-solving
  - Learning
- Adaptive behavior
  - Conceptual Skills – literacy, math, money, time
  - Social Skills – relationships, social judgement
  - Practical skills – self-care, daily living skills, safety

Down syndrome is the most common genetic cause of intellectual disability

Assessment: Cognitive testing (IQ) + Adaptive functioning assessment (ABAS, Vineland)

Autism

Challenges with both:

- Social Communication + Social Interaction
  - Social-emotional reciprocity
  - Nonverbal communication
  - Relationships
- Restricted, repetitive patterns of behavior (2/4)
  - Stereotypies
  - Routine dependence/inflexibility
  - Restricted interests
  - Sensory processing differences

Onset during the developmental period, must cause functional impairment, and not better explained by cognitive limitations

Assessment: ADOS, ADI-R, CARS

How common is Autism in DS?

- DS+ASD → 16-39% of children with DS
  - 10× higher than the general population (~1 in 36)
- Parents are usually the first to have concerns
- Average delay from first parental concern to ASD diagnosis is ~4.5 years
  - **Diagnostic overshadowing**
    - Everything is attributed to DS alone
    - Overlapping behaviors in DS and DS+ASD
  - **Developmental delays –** diagnosis requires judgment of communication/play/etc. relative to overall developmental level
    - May be hard for non-experts
    - May take time until true discrepancy can be determined (often ~18-24 months developmentally)
  - **Few standardized assessment tools** specific to or normed for people with DS

When Something Feels Different…

- Parents of children with DS+ASD often describe a gut feeling:
  - **"My child is different from other kids with Down syndrome"**
- Common early concerns that prompt families to seek evaluation:
  - Repetitive movements (stereotypies) — tend to be preoccupying
  - Severe communication difficulties — beyond what's expected for DS
  - Behavioral challenges — aggression, self-injury, extreme rigidity
  - Social withdrawal — not engaging the way other children with DS typically do

Social Differences

	DS alone	DS + ASD
Social Motivation	Seeks out people and interaction; People >> Objects	May prefer to be alone; less interested in engaging others**; Objects >>People
Eye Contact	Uses eye contact to connect and communicate	Inconsistent or brief; not used to share feelings / communicate
Using Gestures	A strength — points, waves, shows things, often before words develop; compensatory	Fewer gestures; may not point or show things to share interest. Doesn't use readily to compensate for verbal challenges
Sharing ("Joint Attention")	Follows your point; brings things to show you; "checks in" with you; points with eye contact to show and share	Less likely to follow your gaze, show you things, or check in; doesn't point to share interest or attention
Copying Others (Imitation)	Good at imitating actions, gestures, and expressions	Harder time imitating; still often better than kids with autism alone, but noticeably less than other kids with DS
Play	Enjoys playing with others; develops pretend play (dolls, action figures, dress up) over time	May prefer playing alone; may repeat the same action rather than playing pretend (spinning or lining up toys, etc.)
Friendships	Warm, affectionate; enjoys back-and-forth social exchanges	May seem "in their own world"; less back-and-forth interaction, less responsive to social bids from others**
Sharing Emotions	Expressive — shares excitement and joy with others; Notices emotional state of others	Less likely to share enjoyment; expressions may be harder to read; may not respond to emotions of others
Responding to Name	Turns and looks when you call their name (when appropriate for developmental level)	May not consistently respond when their name is called
Communicative Intent	Motivated to communicate using sounds, gestures, and expressions	Less drive to communicate or get your attention; may treat other person like a tool (placing parent's hand on an object)

***Can be less pronounced compared to idiopathic ASD*

References: Moss et al. (2013); Patel et al. (2025); La Valle et al. (2025).

Behavioral & Sensory Red Flags

Sensory Seeking

More repetitive/self-directed, may get stuck in sensory loops, less interest sharing the experience with others; Major differentiator (Baumer 2024)

Sensory Avoidance

More extreme avoidance and less flexible – haircuts, toothbrushing, textures/foods, loud sounds; less habituation over time.

Stereotypies

Intense motor stereotypies (hand flapping, body rocking) significantly elevated over DS-only baseline (Moss 2013).

Restricted Focus

Very restricted, intense interests (staring at lights, spinning wheels, etc.). A key differentiator (Patel 2025).

Note: Ritualistic behavior + routine-dependence are often shared across both groups → may not reliably distinguish between DS and DS+ASD

Focus on Frequency, Intensity, & Functional Impairment!

References: Baumer et al. (2024); Patel et al. (2025); Hamner et al. (2020).

Other Red Flags

Repetitive Play

Repetitive, often dominated by sensory seeking with limited variation, may be cause-effect only without progression toward pretend play

Self-injurious behaviors

50% in DS+ASD compared to ~20% in DS alone

Developmental Regression

Loss of previously acquired social/communications skills, often occurring between ages 3-7 in DS population

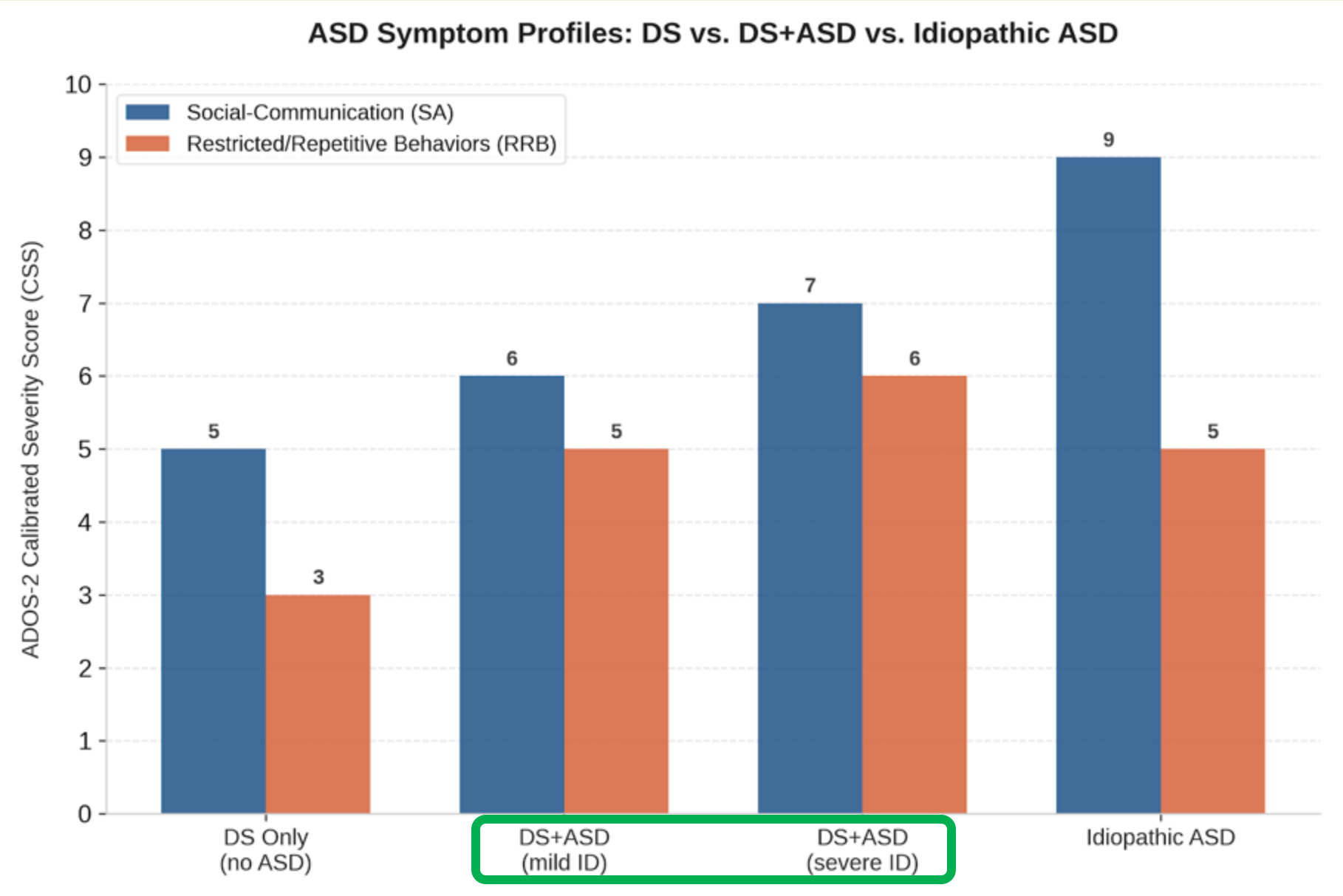
Adaptive Skills

Lowest overall functioning and cognition compared to DS or ASD alone; impaired socialization beyond what IQ predicts (Hamner 2020).

Focus on Frequency, Intensity, & Functional Impairment!

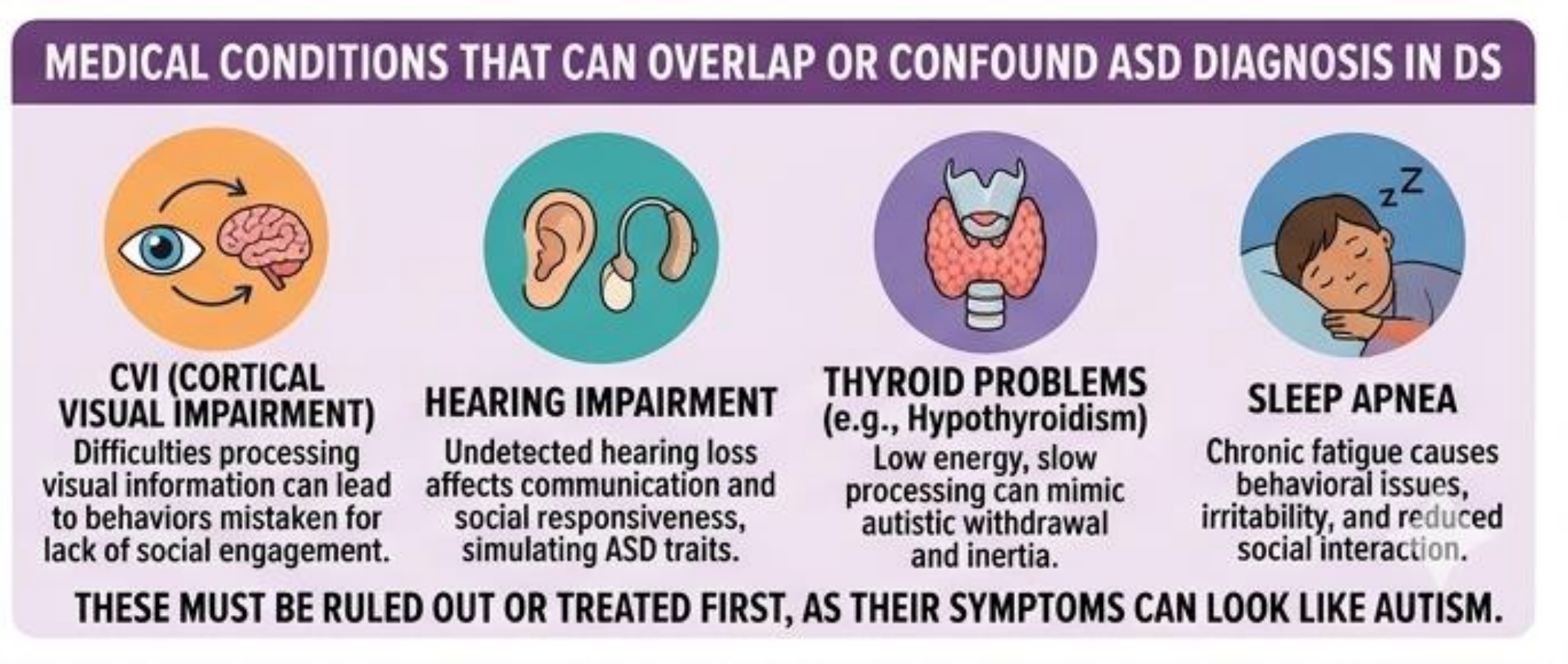
References: Baumer et al. (2024); Patel et al. (2025); Hamner et al. (2020).

Symptom Profiles: DS+ASD is different…



References: Godfery et al. (2019); Hamner et al. (2020).

It Can Get Complicated…



Some conditions that may co-occur more in individuals with DS+ASD and/or can confuse diagnosis:

- **Behavioral sleep disorders**
- **Seizures / epilepsy (infantile spasms)**
- **ADHD / hyperactivity symptoms**
- **GI symptoms – constipation, reflux**
- **Feeding issues**
- **Scoliosis/orthopedic issues?**

Key questions to ask yourself:

Is my child's social engagement and interest in communication consistent with what would be expected for their overall developmental level — or significantly behind?

Are repetitive behaviors, restricted interests, and sensory issues pervasive and intense enough to disrupt everyday life activities and learning?

Have we evaluated/addressed medical issues that may impact behavior?

Then what?... The Autism Evaluation

Who? Someone who knows Autism & DS... typically, developmental-behavioral pediatricians, neurodevelopmental disabilities physicians, neuropsychologists; sometimes primary care physicians or child neurologists.

1. Evaluate for Medical + Neurobehavioral Confounders that may cloud the picture

Hearing loss, vision problems, sleep apnea, and thyroid disease are common in DS and can contribute to symptoms that may look like ASD. Sometimes ADHD, anxiety, or other neurobehavioral conditions may also provide better explanations for the signs and symptoms you may be seeing, or may occur along with ASD.

2. Establish a Developmental Level

Cognitive and adaptive behavior assessments to determine the child's developmental level — so the team knows what social skills to expect. Discrepancies can be harder to identify at lower developmental levels.

3. Parent/School Questionnaire Data

You fill out standardized checklists about your child's social behavior, communication, and repetitive behaviors (e.g., SRS-2, ABC, ND-PROM) – none of these have been validated in DS+ASD (take results with a grain of salt!).

4. Direct Observation (ADOS-2)

The "gold standard" — a 40–60 minute play-based assessment where a trained clinician observes your child's eye contact, gestures, play, and social responses in structured situations. Also not "normed" for DS.

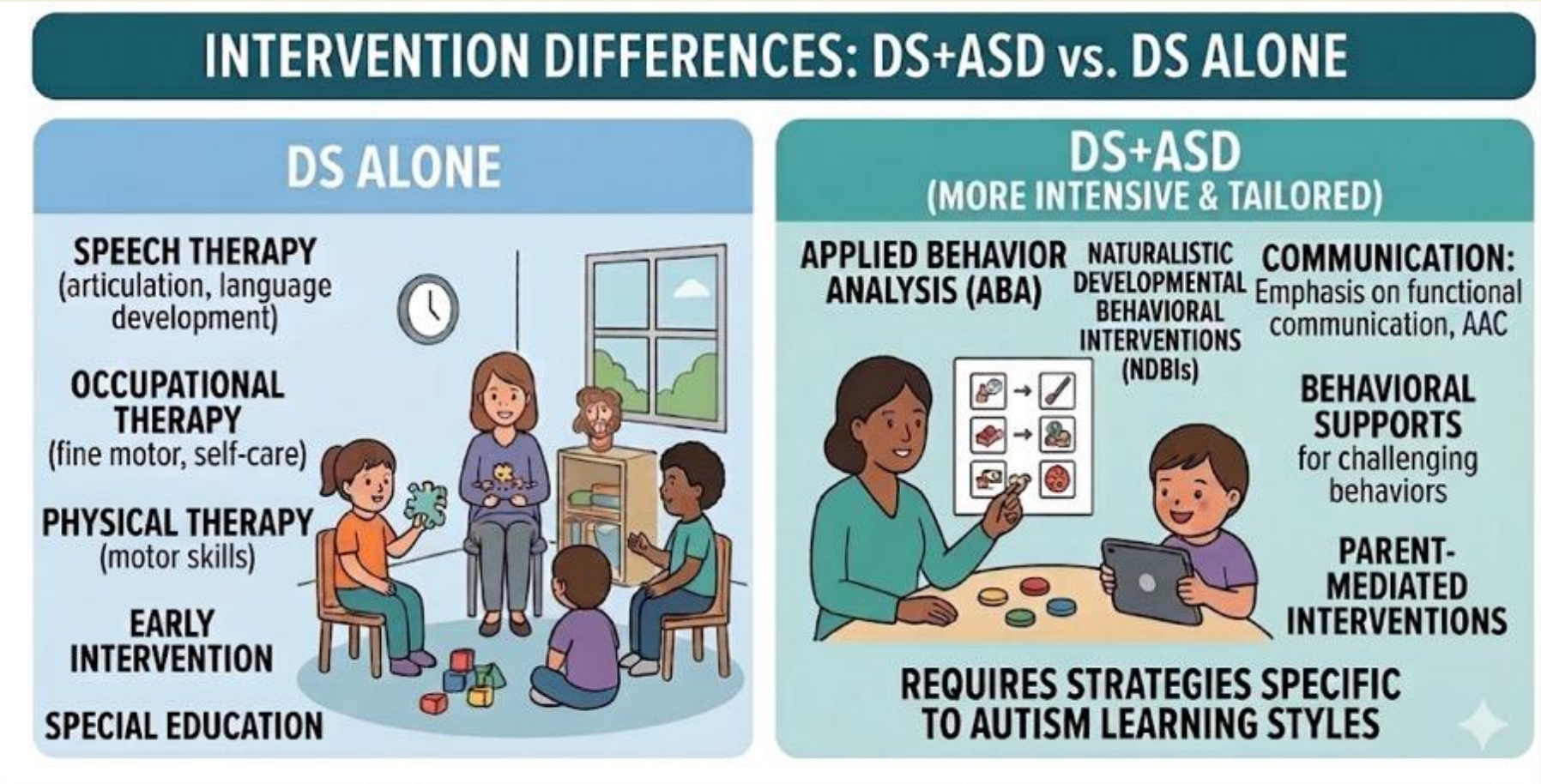
5. Clinical Judgment

No single test gives the answer. The specialist puts it all together — developmental level, observations, questionnaires, ADOS-2 results, etc. — to determine whether autism is present. Some children have such significant cognitive delays that it can be hard to determine if ASD is an accurate diagnosis.

A Quick Aside:
Why might Autism be more common in DS?

- Extra chromosome→ overexpression of genes involved in autism pathways
  - DYRK1A → affects how brain cells grow, connect, and communicate with each other — too much of this gene can disrupt normal brain development
- Imbalance between "go" signals and "stop" signals between nerve cells
  - Associated with autism, seen in DS
- Overactive immune system from birth
  - Inflammation → impact on brain growth/development
- Brain morphology/anatomic differences
  - Cerebellar abnormalities
- Additional genetic differences? (the other 46 chromosomes!)
  - **Role for additional diagnostic genetic testing, like whole exome/genome sequencing?**

Why does it matter?

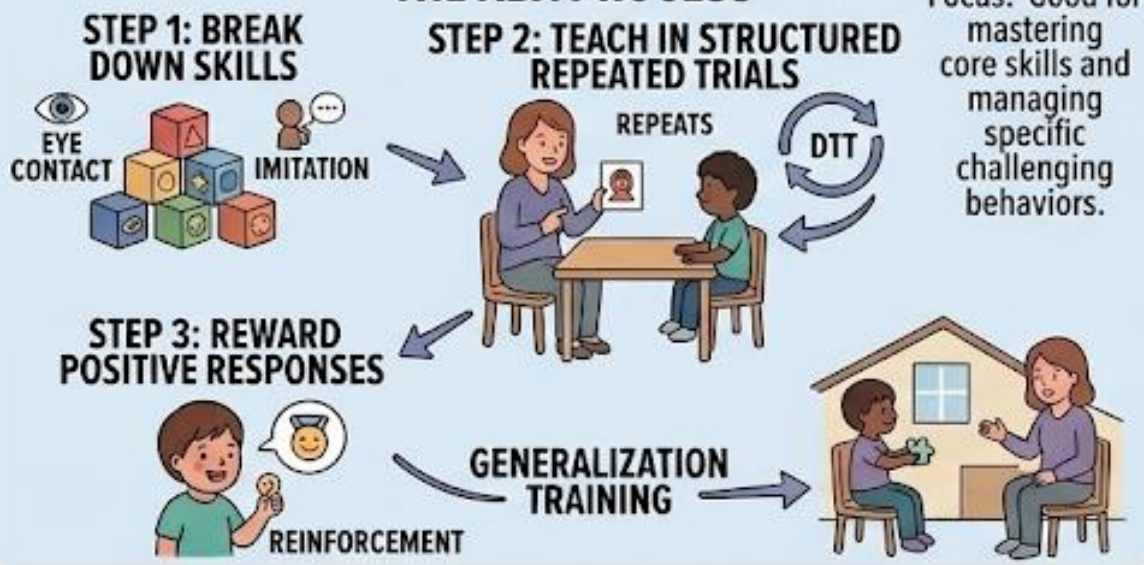


- Improve access to ASD-specific therapies and school supports
- Helps with understanding behavior
- Guides communication + sensory supports
- Can improve family stress and expectations
- Earlier intervention → better outcomes

UNDERSTANDING INTERVENTIONS FOR DS+ASD: ABA vs. NDBIs

APPLIED BEHAVIOR ANALYSIS (ABA)
(SKILL-BUILDING FOUNDATIONS)

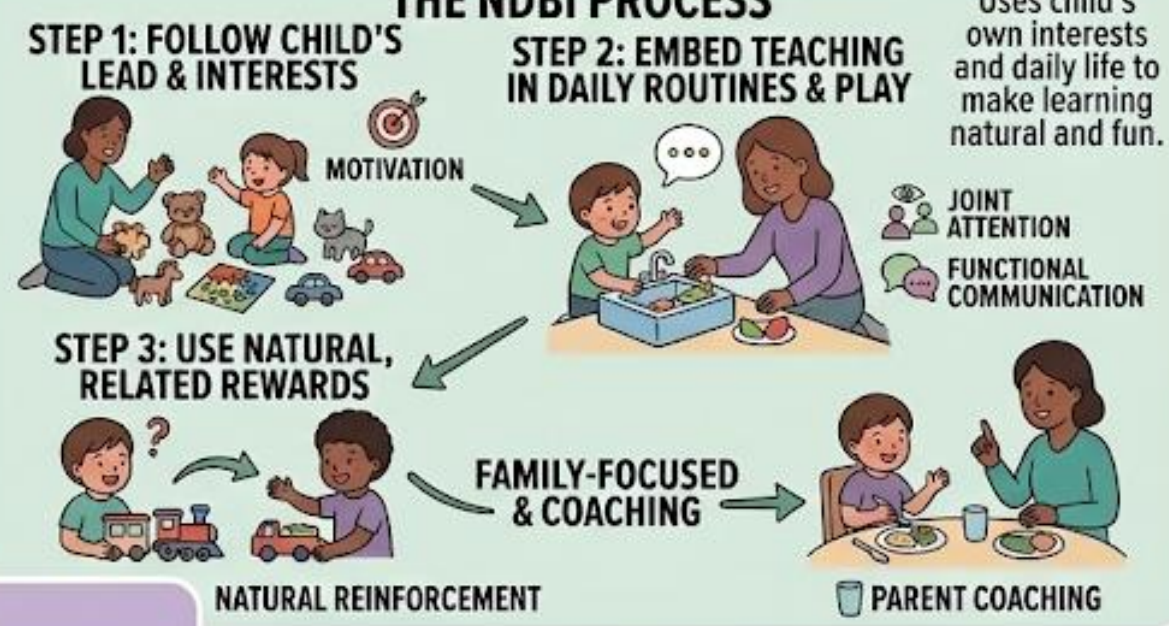
THE ABA PROCESS



Focus: Good for mastering core skills and managing specific challenging behaviors.

NATURALISTIC DEVELOPMENTAL BEHAVIORAL INTERVENTIONS (NDBIs)
(PLAY-BASED, CHILD-LED GROWTH)

THE NDBI PROCESS



Uses child's own interests and daily life to make learning natural and fun.

KEY DIFFERENCES AT A GLANCE

Dimension	ABA	NDBIs
Learning Setting	Structured, often separate area	Natural, everyday environment
Materials	Therapist-selected	Child-selected from choice
Reinforcers (Rewards)	Can be external (e.g., token, treat)	Natural & related (e.g., gets desired toy)
Parent Role	Involved in generalization	Central, active participant

CHOOSING FOR DS+ASD: A BLEND IS OFTEN BEST!

Modern approaches often combine the structure of ABA with the natural warmth of NDBIs. Focus on what is child-centered and joyful for your family!

NDBIs: Naturalistic Developmental Behavioral Interventions

- **JASPER (Joint Attention, Symbolic Play, Engagement, and Regulation)** — play-based naturalistic interactions where adult follows child's lead to create opportunities for shared focus
- **Early Start Denver Model (ESDM)** — combines ABA principles with developmental, relationship-based approaches in naturalistic settings
- **Pivotal Response Treatment (PRT)** — focuses on "pivotal" areas like motivation and self-initiation; by increasing a child's motivation to engage socially, joint attention emerges more naturally

Practical Home Strategies: Behavior

- Visual Supports (learning by seeing >> hearing)
- Simplify language
- Predictable Routines (visual schedules, consistency)
- Prepare for transitions (timers/warnings, social stories)
- Offer choices
- Reduce sensory overload when possible
  - Identify triggers
  - OT with sensory-informed strategies can help
- Teach proactive functional communication
  - AAC: Augmentative and Alternative Communication
- Follow the child's interests to build engagement

Practical Home Strategies: Social Engagement

- Follow your child's lead
  - Join their play
  - Imitate them
  - Create opportunities for back-and-forth
- Build joint attention
  - Point and wait
  - Use animated facial expressions/voice
  - Celebrate when your child shares attention with you
- Predictable social routines
  - Concrete, achievable skills ("hand toy to sibling during play")
  - Practice daily
- Peers
  - Modeling of typical social skills (peers with or without DS)
  - Social skills curriculum (older children)

School Considerations

- “Best” placement varies widely
  - Autism needs may be missed in inclusive settings
  - Is the environment predictable? Are staff trained? Are social expectations scaffolded? Are supports adequate? Is behavior interpreted developmentally?
- Structured with visual supports + predictability
- Social goals → individualized + included in IEP
  - Explicit teaching/modeling of social skills
- Behavior plans should include sensory and communication supports
  - Proactive, not crisis-response only
- Parents often need to advocate for autism-informed accommodations

Other options:

Telehealth and Self-Directed Programs for Caregivers of Children with DS + Autism

Program Category	Program / Option Name	Key Features & Description
Programs with Direct Parent Coaching	Project ImPACT online	Weekly for 6 months
	Telehealth Pivotal Response Treatment (TPRT)	Weekly for 3 months
	Statewide NDBI Coaching	State-dependent, integrated into EI and special education services
	WHO Caregiver Skills Training (CST)	Free, open-access program; 9 group sessions / 3 home visits
	RUBI Parent training	DS-adapted version; Also potential for telehealth delivery
Self-Directed (on-your-own) Programs	Project ImPACT Online	Self-paced version with no live therapist coaching
	Online Parent Training in Early Behavioral Intervention	Complete behavioral intervention training

Medications?

- **No medications improve the core symptoms of autism**
  - Leucovorin → Not generally recommended
    - Poor evidence base; Retracted randomized clinical trial; data based on a handful of small, single-center trials with variable outcome measures and significant conflicts of interest
- Medications can sometimes help with **associated symptoms**
  - Hyperactivity (alpha-agonists, stimulants)
  - Aggression (alpha agonists, atypical antipsychotics)
  - Anxiety (SSRIs)
  - Self-injurious behavior (behavioral interventions! Above meds, naltrexone?)

What parents should do next

Trust	…your observations/instincts
Collect	…examples and videos, information from home and school, etc.
Seek out	…a comprehensive/specialty evaluation
Connect	…with specialized service providers and the DS+ASD community
Focus	…on support and services, not labels

References: Moss et al. (2013); Patel et al. (2025); La Valle et al. (2025).

KEY TAKEAWAYS

UNDERSTANDING AUTISM IN INDIVIDUALS WITH DOWN SYNDROME (DS)

Autism is Not Rare in DS

Affects approximately 1 in 5-10 individuals with DS.

Looks Different than Autism without DS

Flatter symptom profile (social deficits and repetitive behaviors similar in magnitude). Subtler social challenges sometimes.

Diagnostic Overshadowing: A Major Issue

Symptoms are often masked or incorrectly attributed to ID alone.

Diagnosis Opens Doors

Understanding behavior, access to specialized therapies (ABA, NDBI), and parent training resources.

Looks Different than DS Alone

Increased social withdrawal, more challenging social communication, extensive stereotypies and repetitive tendencies.

Powerful Home Tools

Visual supports, predictable routines, alternative communication (AAC), sensory strategies.

Trust Your Instincts: You Are Your Child's Best Advocate!

You know your child best! Ask for support and arm yourself with knowledge!

Future Directions: Possibilities with DS-Connect

- More comprehensive data about DS+ASD
- Recruitment for research studies
  - Evaluation of assessment tools
  - Large-scale validation of Parent Questionnaires
  - Eventual clinical trials
- Potential identification of care gaps across the country
- Linkage of data to the INCLUDE Data Coordinating Center
  - Exploration of genetic, proteomic, neurophysiologic, and other biomarker information
  - Expand understanding of biological basis of DS+ASD

DS-Connect®
The Down Syndrome Registry

Navigating the DS+ASD Journey: Key Resources

The Down Syndrome-Autism Connection™
The only national organization dedicated entirely to DS-ASD.
Features webinars, resource packets, and a private parent support group.

National Down Syndrome Congress (NDSC)
Offers a dedicated online *DS-ASD Guidebook* for families.
Features specialized educational tracks at national conventions.

Down Syndrome Diagnosis Network (DSDN)
Hosts private, age-specific "Dual Diagnosis" parent groups.

Essential Reading
When Down Syndrome and Autism Intersect by M. Froehlke & R. Zaborek.
A New Course by Teresa Unnerstall

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