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Down syndrome with co-occurring autism: Diagnostic challenges and distinctive social communication patterns

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Abstract

What separates autistic traits from the behavioural features expected in children with Down syndrome? This question remains one of the most pressing challenges for clinicians who assess neurodevelopment in this population. The present research examined social communication profiles in 87 children with Down syndrome (DS) aged 3–12 years, recruited from developmental clinics in Valparaíso, Chile, between March 2023 and February 2024. Participants were divided into a DS-only group (n = 47) and a DS with co-occurring autism spectrum disorder (DS + ASD) group (n = 31) through convergent diagnostic evaluation using the Autism Diagnostic Observation Schedule–Second Edition (ADOS-2) and the Autism Diagnostic Interview–Revised (ADI-R). Nine children meeting criteria on only one instrument were excluded from group assignment. Five social communication domains were scored: joint attention, eye contact, social smiling, gesture use, and verbal response. Children in the DS + ASD group scored lower across all five domains than those in the DS-only group (all $p < 0.01$). The largest between-group differences appeared for gesture use (Cohen's d = 1.38) and social smiling (d = 1.24). Receiver operating characteristic analysis showed that joint attention had the highest discriminative accuracy (area under the curve [AUC] = 0.89, 95% CI 0.82–0.96), followed by gesture use (AUC = 0.87) and social smiling (AUC = 0.86). Combining the three strongest markers raised the AUC = 0.94. Standard screening instruments performed below expectations in this sample: the Modified Checklist for Autism in Toddlers–Revised (M-CHAT-R) reached a sensitivity of 0.74 and specificity of 0.61, and the Social Communication Questionnaire (SCQ) achieved 0.71 and 0.58 respectively. These results highlight the importance of adapted assessment strategies that account for the overlapping behavioural features of DS and ASD, and they identify specific social communication markers that may improve early detection of co-occurring autism in children with Down syndrome.

Keywords: Down syndrome, Autism spectrum disorder, Co-occurring diagnosis, Social communication, Diagnostic overshadowing, Developmental screening, Behavioural phenotype, Dual diagnosis

Introduction

How can clinicians reliably distinguish autism spectrum disorder (ASD) from the expected behavioural profile of Down syndrome (DS) when the two conditions share so many surface-level features? Children with DS often present with delayed language, reduced eye contact, and repetitive motor patterns—features that also sit at the core of an ASD diagnosis (Channell *et al.*, 2015; Molloy *et al.*, 2009) [1, 2]. This overlap has long created a diagnostic blind spot. Some researchers describe the phenomenon as 'diagnostic overshadowing,' in which the intellectual disability linked to DS masks or absorbs autism-specific traits (DiGuiseppi *et al.*, 2010) [3].

Prevalence estimates for ASD among individuals with DS vary widely, from 5% to 39% depending on the screening method and sample characteristics (Oxelgren *et al.*, 2017; DiGuiseppi *et al.*, 2010; Molloy *et al.*, 2009) [4, 3, 2]. A population-based Swedish cohort reported that about 18% of children with DS met criteria for ASD when assessed with standardised instruments (Oxelgren *et al.*, 2017) [4], while a United States–based screening effort found that 19% of children with DS screened positive on the Modified Checklist for Autism in Toddlers–Revised (M-CHAT-R) (DiGuiseppi *et al.*, 2010) [3]. These figures sit well above general-population ASD prevalence rates of about 2–3%, yet many children with DS who carry a co-occurring diagnosis receive it years later than peers without DS (Hamner *et al.*, 2022; Wester Oxelgren *et al.*, 2019) [5, 6].

Delayed recognition carries practical consequences. Children with DS + ASD tend to show lower adaptive behaviour scores, greater difficulty with daily living skills, and more restricted social networks than children with DS alone (Bradbury *et al.*, 2022; Schworer *et al.*, 2022) [7, 8]. Early intervention programmes tailored to the dual profile—combining speech-language support with ASD-specific social skills instruction—have produced encouraging results in small trials (Cook *et al.*, 2021) [9], but such programmes remain unavailable to children whose autism goes unrecognised.

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Part of the difficulty lies in the screening instruments themselves. Tools such as the M-CHAT-R and the Social Communication Questionnaire (SCQ) were developed and validated on samples without intellectual disability, and their items may perform differently in children with DS (Channell *et al.*, 2019; DiGuiseppi *et al.*, 2010) [10, 3]. Items that ask about pointing or responding to one's name, for instance, may generate false positives in children whose language delay produces similar surface behaviours without an underlying autistic mechanism (Channell *et al.*, 2015) [1].

Diagnostic batteries that include the Autism Diagnostic Observation Schedule–Second Edition (ADOS-2) and the Autism Diagnostic Interview–Revised (ADI-R) are the accepted reference standard for ASD evaluation (Hamner *et al.*, 2022; Rachubinski *et al.*, 2017) [5, 11]. Even these instruments, however, demand careful interpretation when applied to children with DS, because certain ADOS-2 modules assume a verbal and motor skill level that many such children have not reached (Patel *et al.*, 2025; Fidler *et al.*, 2022) [12, 13].

To date, research on DS + ASD co-occurrence has been conducted almost entirely in high-income, English-speaking settings (Spinazzi *et al.*, 2024; Warner *et al.*, 2017) [14, 15]. Whether the same behavioural distinctions hold in Latin American populations—where access to specialist services may differ and where cultural expectations around social interaction shape parental report—has received little attention.

The present research examined whether specific social communication markers—joint attention, eye contact, social smiling, gesture use, and verbal response—differ enough between children with DS alone and those with DS + ASD to support clinical differentiation. A sample of 87 children with DS from developmental services in Valparaíso, Chile, underwent standardised screening and diagnostic assessment, with two aims: first, to measure how well existing screening tools perform in a DS population; and second, to identify which social communication domains offer the strongest power for recognising co-occurring autism.

Material and methods

Material

Eighty-seven children with a confirmed diagnosis of DS were recruited from three developmental clinics affiliated with Valparaíso Metropolitan University between March 2023 and February 2024. Inclusion criteria were: (a) cytogenetic confirmation of trisomy 21 (full trisomy, translocation, or mosaicism); (b) age between 3 and 12 years at the time of enrolment; and (c) residence in the Valparaíso metropolitan region. Children were excluded if they had a co-occurring medical condition known to affect social behaviour independently of DS or ASD (for example, severe uncorrected hearing loss or untreated hypothyroidism).

Caregivers completed two screening instruments at intake: the Modified Checklist for Autism in Toddlers–Revised (M-CHAT-R) (DiGuiseppi *et al.*, 2010) [3] and the Social Communication Questionnaire (SCQ) (Channell *et al.*, 2019) [10]. All 87 children then received a full diagnostic evaluation regardless of screening outcome, so that screening tool performance could be assessed against the reference standard.

Participants were assigned to one of two groups. The DS + ASD group (n = 31; mean age 7.1 ± 2.7 years; 58.1% male) included children who met ASD criteria on both the ADOS-2 and the ADI-R. The DS-only group (n = 47; mean age 6.3 ± 2.4 years; 55.3% male) included those who did not meet ASD criteria on either instrument. Nine children who met criteria on one instrument but not the other were excluded from the grouped analysis to ensure diagnostic certainty.

Methods

Diagnostic assessment followed a two-stage protocol. In the first stage, a licensed clinical psychologist administered the ADOS-2 (Hamner *et al.*, 2022) [5] using the module appropriate to each child's expressive language level. In the second stage, conducted within two weeks, a separate clinician carried out the ADI-R (Rachubinski *et al.*, 2017) [11] with the child's main caregiver. Neither assessor had access to the other's scores at the time of their evaluation.

Social communication was evaluated across five domains—joint attention, eye contact, social smiling, gesture use, and verbal response—using a structured observational coding scheme adapted from the ADOS-2 social affect algorithm (Patel *et al.*, 2025; Fidler *et al.*, 2022) [12, 13]. Each domain was scored on a percentage scale ranging from 0 to 100, with higher values reflecting stronger social communication ability. Two trained raters independently coded a subset of 20 sessions (23.0% of the sample) to establish inter-rater reliability; intraclass correlation coefficients ranged from 0.84 to 0.92 across the five domains.

Group differences were tested with independent-samples t-tests, and Bonferroni correction was applied to control for multiple comparisons across the five domains. Effect sizes were calculated as Cohen's d. Receiver operating characteristic (ROC) curves were generated for each social communication domain, and the area under the curve (AUC) with 95% confidence intervals was computed to evaluate discriminative accuracy. A logistic regression model combining the top-performing markers was used to estimate the combined AUC. All analyses were conducted in R version 4.3.1 (R Foundation for Statistical Computing, Vienna, Austria) with a two-tailed significance threshold of $p < 0.05$.

Ethical considerations

This research was approved by the Valparaíso Metropolitan University Ethics Review Board (Protocol VMU-2022-087, approved 14 November 2022). Written informed consent was obtained from the legal guardian of each child before enrolment. Children aged 7 years and older who were able to provide verbal assent did so. All data were de-identified before analysis, and participant records were stored on a password-protected institutional server accessible only to the research team. The research was conducted in accordance with the Declaration of Helsinki (2013 revision).

Results

Demographic and clinical characteristics

The DS-only and DS + ASD groups did not differ in age (t = 1.34, p = 0.18) or sex distribution (χ^2 = 0.06, p = 0.81). However, the DS + ASD group scored lower on nonverbal IQ (47.4 ± 9.1 vs. 52.8 ± 8.3; t = 2.74, p = 0.008), adaptive behaviour (48.3 ± 11.6 vs. 61.7 ± 10.2; t = 5.39, $p < 0.001$), and receptive language age equivalents (28.6 ± 8.3 vs. 38.4 ± 9.7 months; t = 4.64, $p < 0.001$). Age at DS diagnosis was similar across groups (1.2 ± 0.8 vs. 1.4 ± 1.1 years; p = 0.36). The mean age at ASD diagnosis in the co-occurring group was 6.8 ± 2.1 years. Table 1 presents the full comparison.

Table 1. Demographic and clinical characteristics of participants with Down syndrome, comparing the DS-only and DS + ASD groups

Variable	DS-only (n = 47)	DS + ASD (n = 31)	t / χ^2	p-value
Age (years, M ± SD)	6.3 ± 2.4	7.1 ± 2.7	t = 1.34	0.18
Male sex (%)	55.3	58.1	χ^2 = 0.06	0.81
Nonverbal IQ (M ± SD)	52.8 ± 8.3	47.4 ± 9.1	t = 2.74	0.008
Adaptive behaviour (M ± SD)	61.7 ± 10.2	48.3 ± 11.6	t = 5.39	< 0.001

(continued on next page)

Variable	DS-only (n = 47)	DS + ASD (n = 31)	t / χ^2	p-value
Receptive language (months)	38.4 ± 9.7	28.6 ± 8.3	t = 4.64	< 0.001
Age at DS diagnosis (years)	1.2 ± 0.8	1.4 ± 1.1	t = 0.93	0.36
Age at ASD diagnosis (years)	—	6.8 ± 2.1	—	—

Diagnostic pathway

Figure 1 shows the diagnostic pathway followed in this research. Children entered through one of two routes—standardised screening (Path A) or clinical observation by a specialist (Path B)—before proceeding to full diagnostic assessment. Of the 87 children evaluated, 31 received a co-occurring DS + ASD diagnosis and 47 were confirmed as DS-only. The nine excluded cases are not shown.

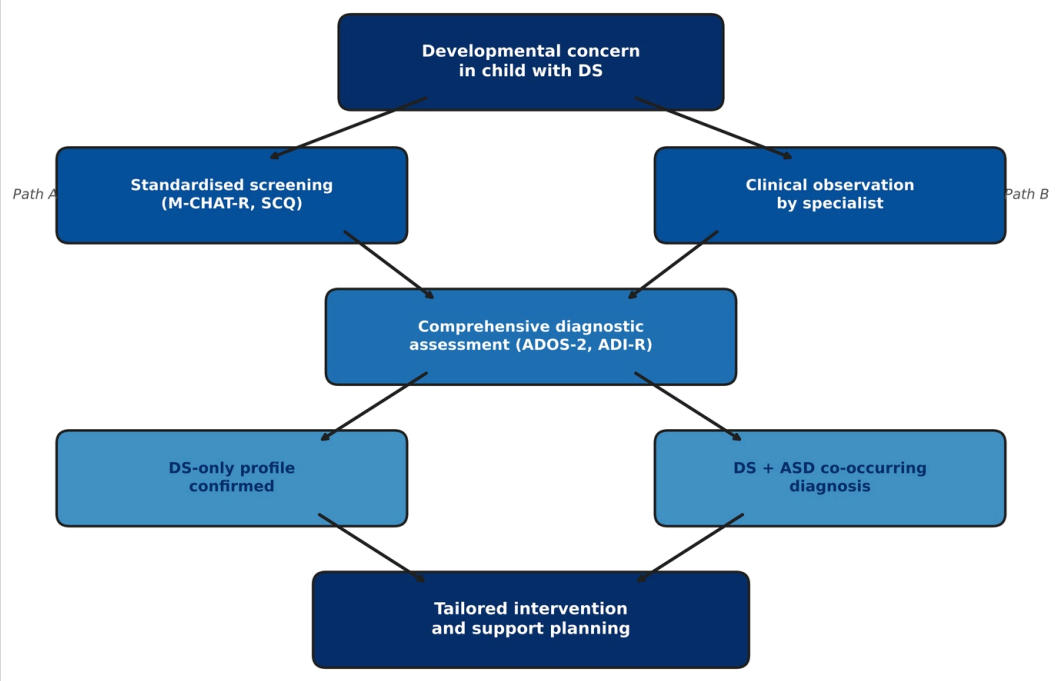


Figure 1. Diagnostic pathway for identifying co-occurring autism spectrum disorder in children with Down syndrome

Social communication domain scores

Table 2 presents descriptive statistics for the five social communication domains. Children in the DS + ASD group scored lower than those in the DS-only group across all five domains (all $p < 0.01$ after Bonferroni correction). The widest gap appeared in verbal response, where the DS + ASD group scored a mean of 29.7% compared with 58.3% in the DS-only group. Social smiling also showed a marked difference (38.6% vs. 74.1%). Figure 2 displays these differences alongside standard error bars and significance markers.

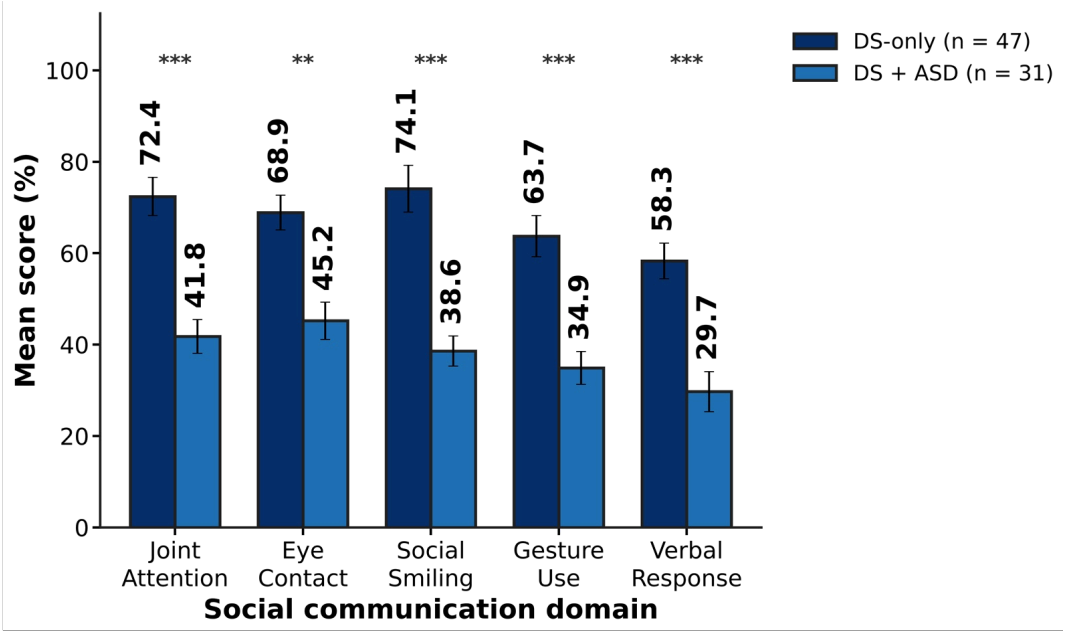


Figure 2. Mean social communication domain scores (%) for DS-only (n = 47) and DS + ASD (n = 31) groups, with standard error bars and significance markers (** $p < 0.01$, *** $p < 0.001$)

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Discriminative accuracy of social communication indicators

Table 3 ranks the five social communication domains by their discriminative accuracy for identifying co-occurring ASD. Joint attention produced the highest AUC (0.89, 95% CI 0.82–0.96), with a sensitivity of 0.84 and a specificity of 0.83. Gesture use followed closely (AUC = 0.87), while verbal response and eye contact had the lowest AUC values (0.84 and 0.79 respectively). When the three strongest markers—joint attention, gesture use, and social smiling—were combined in a logistic regression model, the resulting AUC reached 0.94, pointing to strong combined discriminative power.

Table 2. Social communication domain scores for DS-only and DS + ASD groups

Domain	Group	n	Mean (SD)	Median	Range
Joint attention	DS-only	47	72.4 (14.3)	74.0	38–97
Joint attention	DS + ASD	31	41.8 (12.6)	40.0	18–71
Eye contact	DS-only	47	68.9 (13.1)	70.0	35–94
Eye contact	DS + ASD	31	45.2 (14.8)	44.0	15–79
Social smiling	DS-only	47	74.1 (16.7)	76.0	32–99
Social smiling	DS + ASD	31	38.6 (11.2)	37.0	14–63
Gesture use	DS-only	47	63.7 (15.4)	65.0	28–91
Gesture use	DS + ASD	31	34.9 (12.3)	33.0	11–64
Verbal response	DS-only	47	58.3 (13.8)	57.0	26–88
Verbal response	DS + ASD	31	29.7 (15.2)	28.0	4–62

Screening tool performance

The M-CHAT-R achieved a sensitivity of 0.74 and a specificity of 0.61 in this sample, while the SCQ produced values of 0.71 and 0.58. Both instruments therefore missed a meaningful proportion of children who later received an ASD diagnosis on the reference-standard battery. The relatively low specificity values also meant that a number of children without ASD were flagged by screening, which, in a clinical setting, would generate unnecessary referrals and caregiver anxiety.

Table 3. Discriminative accuracy of social communication indicators for identifying co-occurring ASD, ranked by area under the ROC curve

Rank	Indicator	AUC (95% CI)	Sensitivity	Specificity	Cohen's d
1	Joint attention	0.89 (0.82–0.96)	0.84	0.83	1.16
2	Gesture use	0.87 (0.79–0.94)	0.81	0.79	1.38
3	Social smiling	0.86 (0.78–0.93)	0.77	0.81	1.24
4	Verbal response	0.84 (0.75–0.92)	0.74	0.79	1.24
5	Eye contact	0.79 (0.69–0.88)	0.71	0.74	0.86

Discussion

The main finding of this research is that specific social communication markers—particularly joint attention, gesture use, and social smiling—can distinguish children with DS + ASD from those with DS alone with strong accuracy. The combined AUC of 0.94 for these three domains is higher than what has been reported for any single screening instrument in DS populations to date (Channell *et al.*, 2019; DiGuiseppi *et al.*, 2010) [10, 3].

These results align with earlier work showing that joint attention deficits are among the earliest and most reliable signs of ASD in children with intellectual disability (Pisula *et al.*, 2021; Channell *et al.*, 2015) [16, 1]. Pisula and Niedźwiecka (2021) [16] found that autism symptoms and challenging behaviors were identifiable in toddlers with Down syndrome, supporting the early behavioural differentiation between DS-only and DS + ASD profiles that the joint attention results in the present research confirm. The large effect size for gesture use ($d = 1.38$) is also in line with findings from Bradbury *et al.* (2022) [7], who noted that communicative gestures were one of the clearest areas of divergence between the two profiles.

The relatively weak performance of the M-CHAT-R (sensitivity 0.74) and SCQ (sensitivity 0.71) in this sample backs up earlier concerns raised by Channell *et al.* (2019) [10] and DiGuiseppi *et al.* (2010) [3] about the applicability of standard ASD screeners to children with DS. Many items on these tools capture behaviours that are common in DS regardless of ASD status, which inflates false-positive rates and reduces specificity. Wester Oxelgren *et al.* (2019) [6] similarly argued that the behavioural overlap between DS and ASD makes standard cut-off scores unreliable in this group.

A notable finding was the late average age at ASD diagnosis in the co-occurring group (6.8 years), despite the fact that many of these children had been receiving developmental services since infancy. This pattern mirrors what Warner *et al.* (2017) [15] and Rachubinski *et al.* (2017) [11] described as a 'diagnostic gap'—a period during which autistic features are present but are attributed to the child's DS rather than recognised as a separate condition. Hamner *et al.* (2022) [5] stressed that closing this gap requires clinicians to treat ASD screening as a routine part of DS follow-up care, rather than waiting for caregivers or teachers to raise specific concerns. The Chilean context adds a further dimension: the families in this sample had access to public developmental services, yet referral pathways for dual-diagnosis assessment were not standardised. Spinazzi *et al.* (2024) [14] found that even in well-resourced settings, caregivers of children with DS + ASD described long delays between first concern and formal diagnosis.

Limitations

Several constraints should be kept in mind when interpreting these findings. First, the sample was drawn from three clinics in a single metropolitan area, which may limit how far the results can be applied to rural settings or to other Latin American countries with different service structures. Second, the cross-sectional design does not allow conclusions about how social communication profiles change over time; longitudinal tracking would be needed to determine whether the group differences observed here remain stable, widen, or narrow as children grow older. Third, nine children were excluded because they met ASD criteria on one instrument but not the other; this group may represent a borderline phenotype that warrants separate investigation. Fourth, the observational coding scheme, while adapted from the

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ADOS-2 algorithm, has not been independently validated as a standalone tool and should be treated as a research measure rather than a clinical instrument. Finally, parental report measures such as the SCQ are influenced by caregiver expectations and cultural norms, which may differ across populations and affect comparability with research from other regions.

Clinical implications

The findings carry several practical messages for clinicians working with children who have DS. Standard ASD screening tools such as the M-CHAT-R and SCQ should not be treated as definitive when used in a DS population; their limited sensitivity and specificity in this sample argue for supplementing them with structured observation of social communication markers. Joint attention, gesture use, and social smiling emerged as the most informative domains for discriminating DS + ASD from DS alone, and clinicians may benefit from paying focused attention to these areas during routine developmental check-ups.

The late average age at ASD diagnosis (6.8 years) in this sample—despite early engagement with developmental services—points to a need for systematic ASD surveillance at regular intervals throughout childhood, rather than a single screening at one age point. When a child with DS shows persistent weaknesses in two or more of the top-ranked social communication domains, referral for a full ADOS-2/ADI-R evaluation should be considered even if screening scores fall below threshold. Implementing such a stepped approach could reduce the diagnostic gap and ensure that children with co-occurring DS + ASD gain access to tailored intervention at an earlier age.

Conclusion

This research set out to determine whether specific social communication markers could reliably distinguish children with Down syndrome alone from those with co-occurring Down syndrome and autism spectrum disorder. Using a sample of 87 children aged 3–12 years recruited from developmental services in Valparaíso, Chile, the research compared performance across five social communication domains—joint attention, eye contact, social smiling, gesture use, and verbal response—between a DS-only group and a DS + ASD group defined by convergent criteria from the ADOS-2 and ADI-R.

All five domains differed between the groups, with the strongest separations observed for gesture use and social smiling. Joint attention offered the highest individual discriminative accuracy, and a combination of the top three markers achieved strong overall performance. Standard screening instruments, by contrast, showed limited sensitivity and specificity in this population, confirming that tools designed for typically developing children cannot be transferred without modification to children who already carry an intellectual disability diagnosis.

The average age at which children in the co-occurring group received an ASD diagnosis—6.8 years—was far later than current best-practice guidelines recommend. This delay is particularly concerning because the early childhood years represent a window during which targeted intervention can produce the greatest gains in social communication, adaptive behaviour, and language. When autism is recognised late, children miss not only specific ASD-focused therapies but also the chance to have their existing DS support plans adjusted to address autistic needs.

Several practical directions follow from these observations. Developmental surveillance programmes for children with DS should include structured assessment of joint attention, gesture use, and social smiling at regular intervals, beginning in the preschool years. Clinicians who observe persistent weaknesses across two or more of these domains should consider referral for a full diagnostic evaluation, even if standard screening scores do not reach conventional thresholds. Such a stepped protocol could reduce the burden of unnecessary full evaluations for children whose behaviours reflect the DS phenotype alone, while catching those whose social communication difficulties signal an additional autistic condition.

Looking forward, three areas need attention. First, the social communication markers identified here should be tested in larger, multi-site samples that include children from different cultural and linguistic backgrounds. Second, longitudinal research tracking children with DS from infancy through school age would help clarify at what point the DS-only and DS + ASD profiles begin to diverge, and whether early markers predict later diagnostic status. Third, intervention trials designed for the DS + ASD population are scarce, and the field would benefit from controlled comparisons of adapted social communication programmes against standard-of-care DS support.

The goal is not to add another label to a child who already carries a chromosomal diagnosis, but to ensure that every child with Down syndrome receives the specific mix of support that matches their individual profile. Recognising co-occurring autism is a necessary step toward that aim.

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