Signs of Autism in Infancy: Review Article

A. Shafi[1], S. Raghavan[2], B. Koshy[3], S. Philip Oomen[4]

1. Postdoctoral Fellow, Department of Developmental Pediatrics, Christian Medical College, Vellore, Tamil Nadu
2. Associate Professor, Department of Developmental Pediatrics, Christian Medical College, Vellore, Tamil Nadu
3. Professor, Department of Developmental Pediatrics, Christian Medical College, Vellore, Tamil Nadu
4. Professor and Head, Department of Developmental Pediatrics, Christian Medical College, Vellore, Tamil Nadu

Address for correspondence
Dr. Samuel Philip Oomen
Professor and Head, Department of Developmental Pediatrics, Christian Medical College, Vellore, Tamil Nadu
E-mail: docspo@gmail.com

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Abstract

Background: Autism Spectrum Disorder (ASD) is a neurodevelopmental condition with an early onset, often evident before the age of three. While diagnosis typically occurs later, a growing body of evidence confirms that subtle but significant signs of ASD are frequently present in infancy [.1] Early identification can substantially improve developmental outcomes by facilitating timely intervention during a critical period of brain plasticity.[2]

Objective: This review synthesizes current literature on the early behavioral and neurodevelopmental signs of autism evident in the first year of life, highlighting findings from prospective, retrospective, and high-risk infant sibling studies.

Methods: A narrative review approach was adopted. Peer-reviewed publications between 1990 and 2024 were analysed, including clinical studies, parental report research, video analyses, and prospective cohort studies of high-risk infants.

Results: Key early markers include a decline in eye fixation from 2-6 months[3], limited social engagement and smiling[1], delays in joint attention and gesture use[1], motor delays[1], and atypical affect. Neurobiological studies reveal early brain changes, such as cortical surface area hyper expansion, that precede the full expression of behavioural symptoms.[4]

Conclusion: Infants later diagnosed with ASD often show observable signs within the first year. Recognizing these signs across social, communicative, motor, and sensory domains requires systematic observation and validated tools. Clinicians and early intervention teams must be trained to identify and act upon these subtleties to facilitate timely diagnosis and support.

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1. Neurodevelopmental Basis of Early Autism Signs

The first year of life marks a critical period for brain development, including synaptogenesis, pruning, and network specialization. Disruptions in these processes are increasingly linked to the earliest signs of ASD. [1, 10] Structural MRI studies of high-risk infants have identified a specific developmental cascade: a hyper expansion of the cortical surface area between 6 and 12 months of age, which precedes an overgrowth in total brain volume observed between 12 and 24 months.[4] This rate of brain volume overgrowth in the second year of life has been directly correlated with the severity of social deficits at 24 months.[4]

Functional MRI and EEG studies demonstrate altered connectivity in brain networks, particularly those sub serving face processing and social attention, as early as 6 months of age.[5] Atypical activity in the fusiform face area, amygdala, and superior temporal sulcus has been implicated in the failure of typical eye contact and social gaze development.[5] Furthermore, the genetic underpinnings of ASD are profound, with heritability estimated to be around 80-90%.[6] This risk is polygenic, involving common variants and rare mutations in genes related to synaptic function, such as *SHANK2* and *SHANK3*.[7] Epigenetic mechanisms, like DNA methylation, may link these genetic predispositions to environmental risk factors.[8,9]

2. Review of Methodologies for Early Detection

2.1 Retrospective Video Analysis

Retrospective studies analyzing home videos taken before diagnosis remain valuable for detecting subtle, naturalistic markers. These studies consistently show that infants later diagnosed with ASD exhibit reduced eye gaze, fewer reciprocal smiles, and less imitation compared to typically developing peers. [10]

2.2 Prospective High-Risk Infant Sibling Studies

Infants with an older sibling with ASD have a significantly higher recurrence risk. Prospective studies of these high-risk cohorts provide invaluable insight into the earliest divergence from typical development.[1] By 12 months, siblings later diagnosed with ASD can be distinguished from other infants based on atypical eye contact, visual tracking, and difficulties with disengaging visual attention[1] .Other common early signs in this group include decreased orienting to name, poor visual tracking, reduced motor coordination, and blunted affect by 6–12 months.[1]

2.3 Parental Report and Recall Studies

Parent-reported concerns often precede formal clinical recognition. Parents frequently describe a lack of eye contact, unresponsiveness to their name being called, and limited babbling or use of gestures. Studies have validated that parental concerns, especially when coupled with observational screening, have significant predictive value for a later ASD diagnosis.[11]

3. Core Domains of Early Autism Manifestations

3.1 Social Attention and Eye Contact

While typically developing infants show preferential looking to faces and eyes from birth, a key early sign in ASD is not an initial absence of eye contact, but rather a **decline in eye fixation between 2 and 6 months of age**. [3] This derailment of a fundamental social adaptive process is one of the earliest known indicators of social disability.[3] This pattern is often accompanied by reduced social smiling and poor synchrony with caregivers[1,12]

3.2 Response to Name and Auditory Responsiveness

Failure to consistently respond to one's name by 9–12 months is one of the most robust early markers of ASD.[1,12] This reflects underlying impairments in social orienting and auditory attention, rather than a hearing deficit.

3.3 Joint Attention and Gestural Communication

Joint attention involves both initiating and responding to a shared focus with another person. Deficits in these behaviors, particularly a lack of declarative pointing (pointing to share interest) and showing objects to others, emerge by 12 months in infants with ASD.[1,12]

3.4 Atypical Temperament and Affect Regulation

Infants who later develop ASD are often described as having a blunted affect, low adaptability, heightened negative affect, and poor self-soothing behaviors.[13] These temperamental traits can impact early caregiver-infant bonding.

3.5 Early Language and Vocal Patterns

While some infants with ASD show delayed or absent babbling, others may exhibit more subtle differences, such as producing fewer canonical (consonant-vowel) vocalizations and fewer socially directed sounds by 9-12 months.[14] Unusual crying patterns have also been noted as early as one month of age[5]

3.6 Motor Delays and Unusual Movements

Subtle motor delays are frequently observed alongside social-communicative challenges. These can include poor head control, delays in sitting or pull-to-sit, and atypical reaching and grasping, which may be evident by 6–9 months.[15] Early stereotyped motor movements, such as hand-flapping or body rocking, may also emerge before age one.[16]

3.7 Sensory Processing Atypicalities

Hyper reactivity (e.g., aversion to certain textures or loud noises) and hypo reactivity (e.g., apparent insensitivity to pain, visual fixation on spinning objects) are core features of ASD and are frequently observed in infancy.[17]

3.8 Repetitive and Unusual Play Patterns

By 9-12 months, infants with ASD may show unusual interest in toys, such as atypical visual inspection or repetitive manipulation (e.g., spinning or lining up objects) rather than engaging in functional or symbolic play.[18]

4. Onset Patterns and Developmental Trajectories

The onset of ASD symptoms can follow several patterns:

Early-Onset Pattern: Characterized by clearly observable delays or atypical behaviors before 12 months of age.[19]

Regressive Pattern: Affects up to 30% of children, who lose previously acquired skills—most often language or social interaction—typically between 15 and 24 months.[10]

Plateau Pattern: Children initially meet early milestones but demonstrate a slowing or plateauing of developmental progress after the first year.[20]

5. Predictive Validity of Early Signs

5.1 High-Specificity Markers

Certain signs have very high predictive value for a later ASD diagnosis. These include:

A decline in eye fixation from 2–6 months.[3]

Lack of response to name by 12 months.[12]

Absence of joint attention behaviors (e.g., pointing to share interest) by 12-14 months.[21]

Atypical brain development markers on MRI, such as cortical surface area hyper expansion.[4]

5.2 Multi-Domain Clustering

While individual signs are important, the combination of impairments across multiple domains—social, motor, language, and sensory—significantly improves predictive accuracy.[11,22] A persistent pattern of early atypicalities without developmental catch-up is strongly predictive of ASD.

6. Implications for Clinical Practice

The evidence strongly supports the integration of ASD-specific surveillance into routine pediatric care.

Developmental Surveillance: Paediatricians should actively monitor for the dynamic signs of ASD, such as the trajectory of eye contact and the emergence of joint attention, in all well-baby checks.

Screening Tools: While many screening tools are designed for toddlers (16-30 months), their use highlights the feasibility of early detection. In the Indian context, translated and validated versions

of tools like the **Modified Checklist for Autism in Toddlers, Revised with Follow-up (M-CHAT-R/F)** have shown good psychometric properties.[23] Indigenous tools like the **Indian Autism Screening Questionnaire (IASQ)**[24] and the **Chandigarh Autism Screening Instrument (CASI)**[25] are also valuable resources for community screening.

Parental Concerns: Paediatricians must elicit and validate parental concerns, as they are often the first to notice subtle developmental deviations and are reliable predictors of a later diagnosis.[11,26]

7. Gaps in Literature and Future Directions

Despite significant progress, several gaps remain. There is a critical need for more longitudinal studies that link early signs to long-term outcomes, particularly within diverse populations like India.[27] Further research is required to develop and validate culturally and linguistically appropriate screening tools to improve early detection in low-resource settings.[28] Finally, integrating promising biological markers, such as EEG and neuroimaging, into clinical screening protocols could revolutionize pre-symptomatic identification and intervention.[29]

8. Conclusion

Autism Spectrum Disorder frequently emerges in the first year of life through a constellation of subtle but observable signs affecting social gaze, reciprocity, motor function, and sensory responsiveness. Converging evidence from behavioral and neurobiological research underscores that these are not isolated symptoms but manifestations of an underlying divergence in neurodevelopment. For clinicians, particularly in settings like India where awareness and resources may be limited, a high index of suspicion and a focus on these early markers are paramount. An integrated approach that combines caregiver insight, systematic clinical observation, and the use of validated tools is essential for turning the promise of early detection into a reality for more children.

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