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Review

Early identification of autism spectrum disorders

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HIGHLIGHTS

- There is robust evidence that behavioral signs of ASD can be detected by 1 year.
- Risk markers extend from atypical social communication to motor delays.
- Unusual *trajectories* of language and cognitive skills are reported in ASD.
- A combined behavioral and biomarker approach may help with early detection of ASD.

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ABSTRACT

Earlier identification and diagnosis of autism spectrum disorders (ASDs) can improve opportunities for children to benefit from intervention and lessen the burden on concerned parents. This review summarizes current knowledge about early signs of autism. Convergent data from both retrospective studies and prospective studies of high-risk infants indicate that ASD symptoms emerge in the first two years of life, affecting multiple developmental domains, mapping onto symptom dimensions consistent with current diagnostic frameworks including social-communication, and repetitive interests/behaviors but also extending to motor delays and atypical regulation of attention and emotion. Recent findings have shed new light on patterns of symptom onset and progression, and promise to inform early detection and diagnosis. Further attention to effective application of new findings and related challenges in building health system capacity to ensure timely access to specialized assessment and interventions is needed to fully realize the promise of improved outcomes resulting from this research.

Contents

1. Introduction 134
   1.1. Methodological considerations 134
2. Review findings 134
   2.1. Social-communication 134
      2.1.1. Retrospective studies 134
      2.1.2. Prospective studies 138
   2.2. Language and cognitive development 139
      2.2.1. Retrospective Studies 139
      2.2.2. Prospective studies 139
   2.3. Repetitive interests and behaviors 139
      2.3.1. Retrospective studies 139
      2.3.2. Prospective studies 140
   2.4. Motor development 140
      2.4.1. Retrospective studies 140
      2.4.2. Prospective studies 142

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2.5. Self-regulation and temperament 142
2.5.1. Retrospective studies 142
2.5.2. Prospective studies 142
3. Discussion 143
References 144

1. Introduction

The clinical and etiologic heterogeneity of children with autism spectrum disorders (ASD) contribute to the complex challenges associated with developing a comprehensive early detection strategy. However, it is essential to develop effective approaches to identify and diagnose children with ASD early in life. ASD is one of the most prevalent forms of developmental disability internationally, with current estimates at over 1 in 100 children [1,2]. Earlier diagnosis creates opportunities for children with ASD to benefit more fully from intervention; Dawson's [3] theoretical model goes as far to suggest the possibility of even *preventing* the full manifestations of ASD by taking advantage of early brain plasticity and potentially modifiable abnormalities in reward circuitry early in development [3]. Gains through early intervention can enhance adaptive and cognitive functioning (e.g., [4]) and may ultimately reduce the considerable family and societal costs related to ASD across the lifespan [5,6]. Earlier diagnosis also allows parents to be better informed about recurrence risk to later-born children, and better able to monitor for early signs of autism [7] and other related concerns [8].

This review is aimed at providing a detailed summary of current knowledge of early signs of ASD, from studies across a range of methodologies. Implications for underlying developmental processes and their relation to the emergence of ASD diagnostic features during infancy will be discussed, as will implications for clinical practice and future research.

1.1. Methodological considerations

Research examining early development in ASD has shifted over the past several years from mainly retrospective designs (i.e., through parent report or by examination of early home videos) to prospective longitudinal studies of at-risk infants, generally those with an older sibling with ASD [9]. While retrospective research has generated important insights that have informed current early detection strategies [10], there are some limitations inherent in such study designs. Parental reports of early symptoms of ASD are subject to recall biases; for example, early behaviors more closely related to later manifestations of ASD may be more easily recalled. Pre-diagnostic home videos provide more objective information regarding early behaviors and opportunities for standardized coding, but may be subject to other biases related to sampling. For example, children may be recorded as they are demonstrating a new skill or to commemorate a special event, rather than for the purpose of capturing a more representative range of behaviors. Indeed, the contexts from which behaviors are sampled by home videos vary, both within and across studies, which makes it more difficult to draw conclusions from the literature as a whole [11]. It is important to acknowledge that home video analyses continue to generate important insights; for example, recent studies focusing on behavior less subject to sampling biases (e.g., symmetry of movement [12,13]; see Section 2.4.1 for details) have raised intriguing hypotheses regarding the early motor system in ASD. However, over the past several years, prospective research designs focused on high-risk infants have been increasingly applied to study early development in ASD, with unique methodological advantages. Using this approach, standardized measures can be obtained early in development and over time, generating longitudinal data to map initial trajectories of symptom emergence. Increasingly, behavioral data has been supplemented by experimental measures (e.g., eye tracking, evoked brain responses) which can only be obtained prospectively, thus yielding additional insights regarding underlying developmental processes as well as potential biomarkers that might ultimately contribute to early detection [14]. That said, prospective studies are not without potential methodological limitations. First, affected children from multiple incidence sibships may not be fully representative of all children with ASD. Second, it is difficult to identify comparison groups of infants who are high-risk for developmental delays in prospective research, as risk factors for idiopathic developmental delay (by definition) are difficult to identify, and findings from groups with known risk factors (e.g., Down syndrome) may not generalize to other delayed children. However, prospective studies involving community-referred samples of infants who failed a broad developmental screening have allowed researchers to compare the profiles of infants with ASD to those of infants with other delays.

This review includes a synthesis of published findings that differentiate ASD from typical development and from developmental delay. We summarize these findings by category of behavior (combining early social and communicative features, in accordance with proposed criteria for DSM-5; [15]) and study design (retrospective vs. prospective).

2. Review findings

2.1. Social-communication

2.1.1. Retrospective studies

Retrospective parent report [16–23] and home-video studies [24–39] have found that infants later diagnosed with ASD can be distinguished from infants with typical development (TD) in the first two years of life based on early social-communicative behaviors. Retrospective parental reports, by questionnaire or structured interviews have tended to focus on timing and broad categories of concern (such as speech delay and reduced social-emotional response) and have lacked control groups [16–23]. These studies, mainly published in the 1980s and 1990s, provided powerful evidence that symptoms were present long before many children with ASD were clinically referred, but were generally not designed to characterize specific risk markers. Home video analyses took the field a step forward by using standardized criteria and in more recent studies, incorporating comparison groups of typically developing (TD) and/or developmentally delayed (DD) children, to determine what behavioral features in the first two years of life differentiate infants later diagnosed with ASD (see Table 1 for summary). Coding schemes have progressed from qualitative clinical ratings (e.g., Infant Behavior Summarized Evaluation by Adrien et al. [24]; also see [27,35] in Table 1) to quantitative coding of operationally defined behaviors [25,29,36,40,41].

As summarized in Table 1, several home video analyses have reported that by age 12 months children with ASD can be differentiated from those with typical development (TD) by differences in social communication behaviors. These studies report evidence of reduced/atypical orienting to people, or specifically, people's