

**The Development of Infants with an  
Increased Risk of Autism Spectrum Disorders**

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## ABSTRACT

Previous research has demonstrated that early diagnostic indicators of Autism Spectrum Disorder (ASD) are observable during infancy, often long before a diagnosis is given. Further research was needed to gain greater understanding of the age of symptom onset and the range of deficits and other atypical behaviours. Such knowledge is important for refining early screening and diagnostic instruments, and for targeting early intervention. It is known that individuals who are genetically vulnerable for developing ASD (e.g., siblings of affected children) often present with subclinical features of the disorder, despite not meeting criteria for a diagnosis. Recent research demonstrates that such deviations from typical development may emerge during infancy. This broader expression of the disorder has become widely known as the Broad Autism Phenotype (BAP). A review of the literature shows that early atypical behaviours displayed by both infants later diagnosed with ASD, and those classified as BAP, appear similar. It was thus unclear why similar presentations during infancy resulted in differing diagnostic outcomes.

The present study aimed to further investigate ASD and the BAP during infancy, through a prospective study of early development. The trajectory of development, and early behaviours related to later diagnostic outcome were of particular interest. This study investigated the development of infants who were at-risk for ASD ( $N = 24$ ), and compared this to infants presumed to be typically developing ( $N = 15$ ). Comprehensive assessments of behaviour and development were conducted bimonthly from birth to 18 months of age, with additional assessments conducted at 12, 18 and 24 months of age. A wide range of tools,

including an infant screening tool designed specifically for this study, were administered.

Findings demonstrate that symptomology associated with ASD was observable from early infancy and in some cases from birth. Significant differences between at-risk infants and controls were found on a range of developmental areas and behaviours, throughout the first 18 months of life, including; scores on an infant ASD screening tool, receptive language, cognitive abilities, temperament profiles and sleep behaviours (see Chapter 3).

In total, three children, all at-risk genetically, were diagnosed with ASD at 24 month follow-up. Two of these children presented with atypical behaviours and development from early infancy. A third child appeared to be developing typically to 18 months of age, but experienced a plateau in development and increased stereotypic behaviours between 18 months and 2 years of age. This child was subsequently diagnosed with ASD at 24 months. Case studies (Chapters 4 and 5) of these three children provide insight into early development, and the infant manifestations of ASD.

A more detailed investigation of the group of at-risk infants (Chapter 6) revealed that approximately half of these infants ( $N = 10$ ) presented with symptomology consistent with ASD during early development, but did not meet criteria for a diagnosis of ASD at follow-up. These children were described as presenting with characteristics of the BAP. Five of these BAP children were found to be indistinguishable from those who were subsequently diagnosed with early onset ASD, particularly in the first 12 months of life. Despite a thorough investigation, no specific behaviours emerged that were unique predictors of ASD outcome. Data from this study demonstrate that the prognostic value of these early behaviours was poor

until after 12 months of age. After this age, however, differences emerged in the trajectory of skill development. Those children who presented with early behaviour consistent with ASD, but were not subsequently diagnosed, all experienced a period of dramatic improvement after 12 months of age. Skills including language and cognitive abilities, and performance on ASD screening tools improved considerably. Infants who did not experience this period of 'recovery' were those subsequently diagnosed with ASD. Potential reasons for these divergent trajectories are discussed in Chapters 6 and 8.

Early behaviours that were predictive of later ASD symptom severity were explored (Chapter 7), revealing early skills in social attention most consistently predicted scores on a measures of symptom severity at 18 months of age. The importance of these behaviours for the development of subsequent skills, and collateral impairment that may result from early deficits, are discussed in Chapters 7 and 8.

Overall, the present study demonstrates that behaviours related to ASD are observable during early infancy. Despite similarities in the presentation of ASD and the BAP during early infancy, several children who presented with early indicators were subsequently diagnosed with a disorder on the autism spectrum. Therefore, the ability to recognise symptomology during infancy may allow intervention to commence during the first year of life, potentially altering the trajectory of ASD and reducing the severity of collateral impairments.