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The Puzzle of Autism Spectrum Disorder

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The Puzzle of Autism Spectrum Disorder

Franklin Trimm, MD

INTRODUCTION

Autism is a severely incapacitating and lifelong developmental disorder commonly described as consisting of a triad of impairments in socialization, communication, and imaginative play. Autism was once viewed by health care providers as a condition they were unlikely to encounter during their practice experience. Medical school and residency curriculums often devoted minimal time to addressing autism. There were no uniform methods taught for how to evaluate and manage a child whose development and behaviors suggested autism. Rather, these concerns were felt to be primarily the domain of educational, psychiatric, and psychology professionals.

Autism is now categorized as one of several pervasive developmental disorders (PDDs). Owing to the variations with which autism can present among individuals, as well as the similarities among several of the PDD diagnoses, these entities are sometimes referred to collectively as autism spectrum disorders (ASD). Other PDDs included in this category are Asperger syndrome and PDD–not otherwise specified (PDD-NOS). With no single specific cause known and limited treatment options, autism continues to perplex families and professionals alike.

ETIOLOGY

Approximately 90% of children with ASD do not have a specific etiology identifiable and are classified as having idiopathic autism.¹ As with many developmental conditions, idiopathic autism likely has a multifactorial basis related to the interaction of both genetic and environmental influences. Approximately 10% of ASD cases are associated with or caused by known genetic or medical disorders.² **Table 1** lists a number of syndromes that are associated with ASD. Several perinatal factors have been identified as associated with later identification of ASD and include daily smoking in early pregnancy, maternal birth outside of Europe and North America, cesarean delivery, being small for gestational age, a 5-minute Apgar score below 7, and congenital malformations.³

EPIDEMIOLOGY

Although monitoring the presence of a behaviorally defined disorder can be difficult, recent reports in-

dicating that the prevalence of ASD is on the rise. Surveys of children and adolescents performed prior to 1988 estimated autism to have a prevalence of 4 to 5 cases per 10,000.⁴ Using current diagnostic criteria and approaches, more recent studies report prevalence rates for autism of 21 to 31 cases per 10,000⁵⁻⁷ and of 57.9 cases per 10,000 for all PDDs combined.⁶ The reported increases are likely due to a combination of factors, including changes in diagnostic criteria and increased public and professional awareness, in addition to an actual increase. With this increase in prevalence, the probability of a health care professional encountering a child with autism also increases. In California, the number of children receiving services for autism rose 273% from 1987 to 1998.⁸

The age at which children are diagnosed is dependent on the severity of the disorder and how disruptive the child's behaviors are. Those with more severe symptoms are recognized earlier. Although just a decade ago, the average age at diagnosis was 4 to 6 years, currently it is around 30 months.⁴ Recognizing ASD earlier has a number of benefits. Family stress related to the child's developmental problems can be better managed when a diagnosis and related prognoses can be presented and discussed. Appropriate referral to early intervention services improves the outcome for many children with ASD, particularly when begun early and continued consistently.⁹⁻¹¹ The increasing prevalence of ASD and the need to identify children at a young age to enhance the benefits of early intervention increase the need for health care providers to be knowledgeable and competent at eliciting parental concerns about their child's development, recognizing ASD symptoms, and performing initial assessments.

CASE PRESENTATION

A 36-month-old boy presents for his first visit to a new primary care physician. He is accompanied by his mother, who serves as the historian. She reports that she first had concerns about her son at around 18 months of age. At that time, he was not saying any words that others could understand, he was easily frustrated with subsequent extended temper tantrums, he would not fall