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AUTISM

Why the rise in rates?

Our improved understanding of the disorder and increasingly sensitive diagnostic tools are playing a role—but so are some other factors.

For many years, articles about autism cited prevalence rates of approximately 7 in 10,000.¹ Over the past few years, however, there appears to have been an explosion in the rate at which autism is diagnosed: More recent estimates range from about 30 in 10,000² to one in 68.³ References to an autism epidemic appear to have originated in a 2002 California legislative report suggesting a 273% increase in autism from 1987 to 1998.⁴

Concerns about rising rates of autism, however, are not new. In 1943, Leo Kanner, MD, a psychiatrist and pioneer in the study of autism, published a paper titled, “Autistic disturbances of affective contact.”⁵ The result? “Almost overnight, the country seemed to be populated by a multitude of autistic children,” he later observed.⁶

To what should we attribute the current rise in reported autism rates? Even a casual review of the autism literature suggests a number of potential causes that may account for at least a portion of the recent increase.

We know more about the disorder

In his paper, Kanner described the “pecu-

liarities” of 11 children whom he had cared for.⁵ While several had been diagnosed with mental retardation, childhood schizophrenia, or both, what stood out to Kanner was an “autistic aloneness” evident from the beginning of life. This was in contrast to childhood schizophrenia, in which a child experienced a *departure* from previously normal interrelations. His insight contributed to our understanding of what is now recognized as autism spectrum disorder (ASD).

Also in 1943, Hans Asperger, MD, was studying families with children exhibiting behaviors similar to those described by Kanner. The following year, Asperger published an article (in German) describing these children. Unfortunately, this paper—titled “Autistic psychopathy in childhood”⁷—was not translated into English until the early 1990s.⁸

Since Kanner and Asperger first called attention to the disorder, there have been numerous changes in societal and medical understanding of autism. Bruno Bettelheim, PhD, an Austrian-born child psychologist with a particular interest in emotionally disturbed children, theorized that poor maternal bonding and lack of maternal affection were responsible for autistic characteris-

tics.⁹ Bernard Rimland, PhD, a psychologist and the father of an autistic child, argued for a biological basis of the disorder.¹⁰ Rimland's theory of autism as a neurodevelopmental disorder with an unidentified organic etiology is most consistent with current medical opinion.

Definitions and diagnostic criteria have evolved

The first edition of the *Diagnostic and Statistical Manual of Mental Disorders (DSM)* was published in 1952.¹¹ Although it was nearly a decade after Dr. Kanner clearly described autism as an entity separate from childhood schizophrenia, the word autism was used in this edition only once—to describe psychotic reactions associated with schizophrenia. Similarly, autism was referred to only in relation to childhood schizophrenia in the *DSM-II*,¹² published in 1968.

■ **DSM-III adds diagnostic criteria.** In *DSM-III* (1980),¹³ specific diagnostic criteria for infantile autism and pervasive developmental disorder (PDD) appeared for the first time. Both diagnoses were clearly contrasted with a diagnosis of schizophrenia, and were used to identify children who exhibited a pervasive lack of responsiveness to others.

The *DSM-III-Revised (R)* (1987)¹⁴ added a classification scheme more consistent with the current standard. It included: 1) qualitative impairment in reciprocal social interaction; 2) qualitative impairment in communication and imaginative activity; and 3) restricted activity/interests.

■ **PDD Not Otherwise Specified** was also included, and served to identify those who had qualitative impairments in social interaction and communication skills but did not meet the full criteria for autism disorder or PDD.

The publication of *DSM-IV* (1994)¹⁵ brought another change: Specific criteria were outlined for the diagnoses of Asperger syndrome, Rett syndrome, and childhood disintegrative disorders; in the *DSM-IV-Text Revision (TR)* (2000),¹⁶ this structure remained relatively stable.

DSM-5 (2013) took another step, consolidating these various disorders into a single,

unifying diagnosis of ASD.¹⁷ Significant controversy surrounded this change, with some viewing it as an oversimplification that does not accurately reflect important distinctions among divergent disorders¹⁸ and others arguing that it will result in unrecognized cases and exclusion of affected individuals.¹⁹ Notably, a recent study using concurrent *DSM-IV* and *DSM-5* criteria to diagnose autism and PDD in more than 4000 children documented a high level of agreement between them.²⁰

Diagnostic tools have improved

As the number of children diagnosed with autism has increased, so have efforts to more accurately diagnose autism as a distinct disorder.

In the 1960s, guidelines for diagnosing autism focused primarily on Kanner's original descriptive criteria. Even in the 1980s, after *DSM-III* criteria identified autism as a distinct disorder, children were being evaluated with generalized developmental screening tools focused on behaviors characteristic of a severe mental handicap, without differentiating between autistic and nonautistic children.

Early autism-specific observational and structured interview tools (eg, Childhood Autism Rating Scale,²¹ Autism Diagnostic Interview [ADI],²² and Autistic Diagnostic Observation Schedule)²³ emerged from a need for standardized diagnostic instruments that were comparable and reproducible.²⁴ But because these tools were highly specific and initially studied in research settings with high-risk populations, they lacked the sensitivity to identify children at risk in the general population, particularly those with milder symptoms.

As the diagnosis of autism became more standardized following publication of the *DSM III-R* in 1987, developmental specialists were able to construct increasingly sensitive evaluation tools. The ADI was revised²⁵ to facilitate earlier and more efficient diagnosis, allowing for assessment of children as young as 19 months of age. The Checklist for Autism in Toddlers (CHAT)²⁶ and subsequent modification (M-CHAT)²⁷ were among the earliest and most effective screening tools, appropri-



A blood test to identify differences in gene expression between children with and without ASD has shown initial promise—particularly in males.

Recent studies suggest that advanced paternal age can increase the risk of autism.

ate for use in children as young as 16 months old.

Tools that followed the original CHAT (eg, the Autism Spectrum Screening Questionnaire [ASSQ]²⁸) were adapted to better identify high-functioning children with Asperger syndrome, as well as those with autism.

Another revision of the M-CHAT—the M-CHAT-R/F (Revised with Follow-up) was validated earlier this year. In a study involving 16,000 children, 95% of those who had positive tests were found to have some form of developmental delay and almost half (47%) received an ASD diagnosis.²⁹

■ **Other diagnostic aids** are being explored as a means of promoting earlier identification of ASD. For example, a blood test to identify differences in gene expression between children with and without ASD³⁰ has shown initial promise, particularly in males. This test is licensed by SynapDx (Lexington, Mass) and a clinical trial to evaluate it has begun.

Results of another study demonstrating normalization of brain activity in autistic children after they've undergone intensive treatment³¹ raise the possibility of using cortical activation as measured by electroencephalography as an early biomarker for autism.

Treatment options, advocacy affect rate of diagnosis

Improvements in diagnosis and targeted identification of potentially treatable symptoms³² led to the development of new treatment options. And greater use of day care and preschool programs prompted networking among parents, who touted the benefits of early evaluation, diagnosis, and treatment. Earlier screening, not surprisingly, led to an increase in the target population.

Local as well as national advocacy groups, led primarily by parents, have become powerful voices for improvements in services offered to children with autism. And research studies with varying degrees of sophistication continue to be published, further fueling the demand for school systems to provide supportive learning environments as required by the Individuals with Disabilities

Education Act, initially enacted in 1975 and amended in 2004.³³

Federal and state funds in the form of Medicaid waivers are available to provide long-term care services in home and community settings, while private insurers typically pay for associated treatment modalities for those with an autism diagnosis, including physical, occupational, and speech therapy, among other services.

Finally, politicians and celebrities with personal connections to autism have joined the effort to increase awareness and improve the quality and availability of services—further assuring that autism is recognized as a legitimate, definable, and treatable disorder.

As an autism diagnosis has become more socially acceptable, it has at times replaced diagnoses of learning disability and mental retardation, a trend known as “diagnostic substitution.”³⁴ Indeed, having a child with an ASD diagnosis often makes it possible for parents to secure services that might otherwise be unavailable to them.

Is the incidence of autism linked to the environment?

Numerous environmental, nutritional, and pharmaceutical changes have been cited as reasons for what is perceived as an increasing incidence of autism in recent years. For example, some contend that greater use of food preservatives and greater exposure of young children to environmental toxins are contributing factors.³⁵

■ **Thimerosal.** Perhaps most notable is the assertion—since disproven—that thimerosal, a substance previously used in the manufacture of several childhood vaccines, was a leading cause of autism.^{36,37} In fact, one study documented an *increase* in autism after thimerosal had been discontinued.³⁸ (For more on the thimerosal controversy, see “Autism: 5 misconceptions that can complicate care” on page 310.)

■ **Autism comorbidity.** As autism is frequently comorbid with other developmental disabilities, advances in medical technology that have led to a decline in neonatal death and overall mortality among the disabled may mean more survivors are subsequently diagnosed with an autism comorbidity.

■ **Biological factors.** Recent studies suggest that advanced paternal age can increase the risk of autism.³⁹ Twin studies suggest moderate genetic heritability, along with a substantial environmental contribution to the development of autism.⁴⁰ And new research suggests that maternal stressors during pregnancy—eg, trauma, illness, or substance abuse—may increase a child’s risk of developing autism, among other psychiatric disorders.⁴¹

The important role you play in the diagnosis of autism

It is clear that autism is more common than previously thought³ and that various factors are at work. Ensuring that children are promptly and properly evaluated begins when primary care physicians take parents’ concerns seriously and keep an eye out for common symptoms and characteristic developmental delays that may be evident even in the first year of life.

If symptoms are not severe enough to be detected in the child’s first several years, the next likely presentation will be when a parent gets a call from school suggesting that their child be tested for autism. While this delayed presentation suggests a higher level of functioning, a full evaluation, including the use of questionnaires such as the Social Responsiveness Scale⁴² and ASSQ,²⁸ and appropriate referrals are still vital.

Puberty is another time when characteristics suggestive of autism that escaped earlier detection may be noted, and vague behavioral issues or concerns about intellectual impairment may become sufficiently troublesome to prompt a thorough evaluation. While behavioral therapy can be very helpful in such cases,

TABLE

Autism spectrum disorder: Where to learn more

The Arc (formerly the Association of Retarded Citizens): www.thearc.org
Autism Central (in Canada) http://www.autismcentral.ca/new/index.php/en/research/best-practises
Autism Research Institute* www.autism.com
Autism Society www.autism-society.org
National Autism Association† http://nationalautismassociation.org

* This group provides information about complementary and alternative treatments.

† Complete list of state chapters can be found here.

medical therapy directed at co-occurring conditions (ie, a mood or anxiety disorder, including obsessive-compulsive disorder) may also provide significant benefit.

Whatever the age of the child, his or her parents should be counseled as to the general nature of autism, reassured of the availability of treatment options and given the appropriate referrals, and encouraged to learn more by availing themselves of resources (TABLE) and support groups. A consumer update from the US Food and Drug Administration titled “Beware of false or misleading claims for treating autism” (<http://www.fda.gov/ForConsumers/ConsumerUpdates/ucm394757.htm>) will be helpful for many parents, as well. **JFP**

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