



Autism: A Spectrum Disorder

Recently, during a rotation as the attending physician on our cardiology consult service, a patient we were seeing for atrial fibrillation with rapid ventricular response was reported to me as being autistic. Thinking about this term and the patient being examined, I realized that I really knew very little about autism, and decided it was time to do some research on this entity. What I discovered was, in fact, quite interesting.^{1–5} I am writing this commentary because autism is common here in the United States, and because I believe that it is important for internists to know more about this condition, which can affect the care of individuals with other illnesses commonly seen by internal medicine physicians.

Let's start with a definition of this entity. Autism is a spectrum disorder, that is, it presents with a broad array of severity. In its most severe form, this disease can completely incapacitate an individual who will then require life-long supportive care often in a chronic healthcare institution. In its milder form, autistic individuals can lead normal, rich, and productive lives having learned coping strategies. Autism is defined in the psychiatric literature as a neurodevelopmental disorder characterized by failure on the part of the affected person to communicate and interact socially with others. Autistic patients commonly demonstrate restricted, repetitive, and stereotyped patterns of behavior. For example, as children or adults, autistic individuals may continue to repetitively handle and play with objects such as toy automobiles rather than interacting with other persons. These repetitive actions are referred to as "stimming"—self-stimulating behaviors, usually involving repetitive movements or sounds. Autism is found in individuals throughout the world and has no specific propensity for any race, culture, or economic status. It is 4 times more common in males than in females and is usually diagnosed in childhood when parents and teachers observe that the affected individual fails to make eye contact and interact normally with others. The Centers for Disease Control and

Prevention have estimated that 1 in 59 children are autistic and since this is not a fatal condition, the number of adults with autism is substantial.

The *Diagnostic and Statistical Manual of Mental Disorders* published by the American Psychiatric Association contains criteria that enable clinicians to diagnose a variety of psychiatric disorders including autism. In this manual, 5 different autism subtypes are recognized: autism with or without intellectual impairment; autism with or without language impairment; autism accompanying another medical or genetic condition; autism associated with another neurodevelopmental, mental, or behavioral disorder; and autism combined with catatonia. Adding to the complexity, individuals with autism can simultaneously exhibit elements from more than one of these subtypes. In the past, individuals with autism may have been diagnosed with entities such as Asperger's syndrome or some form of developmental delay. Currently, however, these latter diagnoses are not used. Rather, as noted above, patients are diagnosed as being on the autism spectrum.

The etiology of autism is not known at this time. However, a number of risk factors have been identified, including familial inheritance; genetic mutations leading to abnormalities of brain structure or function; birth to older parents; low birth weight; exposure to heavy metals, environmental toxins, or both; viral infections affecting the central nervous system; and fetal exposure to specific medicines, such as valproic acid and thalidomide. Authorities at the National Institute of Neurological Disorders and Stroke believe that both genetics and environment play a role in the etiology of autism. A common misconception is that some forms of vaccination can cause autism. This is simply not true, and the work that led to this idea has been retracted.^{6,7} The diagnosis of autism is based on clinical observation; no biomarkers have yet been found to assist with making the diagnosis, which is almost always made during childhood. Genomic screening is helpful in some individuals with known genetic diseases. The diagnosis usually requires evaluation by a team of specialists including psychologists, occupational therapists, and speech-language pathologists.⁸

What therapies are available for individuals diagnosed with autism? Because the entity is usually identified in childhood, a variety of pediatric psychological therapies

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have been recommended, including behavioral, play, and occupational therapy alongside physical and speech therapy. Massage is recommended by some to induce relaxation in individuals who are hyperactive. For adults, behavioral and physical therapy may be continued, but many individuals with autism have learned coping strategies by the time they reach adulthood. In addition, many adults with milder or moderate forms of the spectrum can lead relatively normal, productive lives. A number of alternative therapies have been suggested such as chelation. However, none of these has been definitively shown to help. Some authorities believe that diet may play a role in improving symptoms in individuals with autism spectrum. A variety of diets have been employed, including limiting foods with artificial additives or gluten while emphasizing fresh fruits and vegetables, lean meats, and fish, and carrying out efforts to maintain hydration. Research concerning dietary therapy is still inconclusive at this time.⁸ Others have suggested regular exercise as a form of therapy for these patients, which is likely to offer benefits even if it does not cure them.

Whether adult patients with the autism spectrum can lead relatively normal lives depends on the severity of the expression of the disease and whether as children or adults they have responded to some or all of the therapies listed above. Patients with milder forms of the spectrum may not be diagnosed until they reach adulthood. The therapies listed above may be helpful in controlling some aspects of the spectrum even when initiated for the first time in an adult patient. Sometimes individuals who are on the spectrum are first diagnosed much later in life. This is due in part to a lack of awareness among medical practitioners, and hence this commentary. Some confusion exists about whether attention deficit disorder (ADHD), a common entity, is part of the autism spectrum. Many clinicians who deal with this latter group of patients do not believe that ADHD is part of the autism spectrum because ADHD patients do not usually lack socio-communicative skills. Prognosis for individuals with autism generally depends on how early in life the spectrum was identified, the severity of

disability, and the effect of the various therapies listed above. There is no cure for autism, but the spectrum is not lethal. However, many patients are unable to function normally in society. Management of comorbid disease can be challenging in adult patients with autism because of failure to adhere to guideline directed therapies.

As always, I am happy to hear from readers concerning this commentary at jalpert@shc.arizona.edu.

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