

# Expanding Our Knowledge of Autism

By Jack M. Gorman, MD

In our quest to find topics of general interest to both psychiatrists and neurologists, we are beginning the new year with a perfect subject: autism. Patients with autism are typically evaluated by both child psychiatrists and pediatric neurologists, and both disciplines have contributed importantly to the ongoing dialogue about the underlying pathophysiology of this serious and often tragic brain disease.

Several new ideas about autism are currently emerging. First, the disorder seems to be far more common than previously believed, at least insofar as "spectrum" diagnoses are included in overall prevalence rates. This was originally ascribed to better detection and more systematic epidemiology. While that is certainly a contributing factor, it is possible that the incidence of autism is indeed increasing. For example, I was recently told that the incidence of autism has increased dramatically in California over a period of just a few years; much too quickly to be simply ascribed to better recognition of the illness.

Second, it is increasingly apparent that autism is in part a genetic disease. Family studies have been leading us to this conclusion for years. At the recent annual meeting of the American College of Neuropsychopharmacology, Mount Sinai School of Medicine neuroscientist Joseph Buxbaum, PhD, announced the discovery of strong evidence for a specific gene contributing to the risk for autism. Buxbaum and colleagues, along with other researchers, also have leads on several other risk genes, suggesting that similar to Alzheimer's disease and breast cancer, autism will turn out to be linked to a set of vulnerability genes that, taken together, increase the chances of disease acquisition. Whether there are specific environmental factors that add to this risk is yet to be determined, although many putative risks, such as childhood vaccinations, seem to have been dismissed by scientists. More on this subject is sure to follow.

Third, abnormalities in brain activation have been consistently reported in autism. These include parts of

the brain that one might predict would be related to the condition, such as the prefrontal cortex and amygdala, and areas that at first glance are not so obvious, such as the cerebellum. One of the current limitations in the neuroimaging area that most studies involve adults with autism or syndromes along the autism spectrum. Cognitive neuroscientists now know just how much plasticity there is to the human brain; what we see in an adult brain represents years of learning, experience, and compensation for congenital deficits. Hence, there is a real need to perform extensive neuroimaging studies on children with autism if we are going to understand the basic neurocircuitry abnormalities. This is not easy. Some neuroimaging methods are currently considered inappropriate for research with minors because, like positron emission tomography, they involve exposure to low levels of ionizing radiation. This leaves the various applications of magnetic resonance imaging which, although likely to be highly revealing, require that a subject remains still in a small tube surrounded by loud noises for prolonged periods of time. This work is a challenge but can be done, and scientists on my team and others are now pushing ahead with their plans.

Eric Hollander, MD, has emerged as a national leader in autism research, having assembled a terrific team of basic and clinical scientists to study genetics, imaging, and treatment options. We are grateful to him for taking the time to put together this group of outstanding review papers that will hopefully help guide scientists, clinicians, and families in the struggle to conquer autism.

In closing, I want to take this opportunity to wish all of our 50,000 readers a wonderful, healthy, and Happy New Year. *CNS Spectrums* enjoyed great success in 2003 and we have plans to build upon that success in the new year. We continue to welcome your manuscripts, case reports, letters to the editor, and suggestions. More than anything else, we want you, on every level, to feel involved because you are an integral part of the *CNS Spectrums* family. **CNS**

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