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The Autism Spectrum

by Simon Baron-Cohen

In a one-on-one session, a therapist works with a five-year-old boy with autism to improve his cognitive skills. During their sessions the therapist subtly switches between play and learning.

In 2009 researchers made numerous discoveries concerning the prevalence, neuropathology, and treatment of autism spectrum conditions (also known as autism spectrum disorders). Indeed, new estimates of prevalence in both the United States and the United Kingdom indicated that roughly 1 in every 100 children was diagnosed with one of these conditions. Autism is a spectrum condition—it is manifested to varying degrees of severity. At one extreme a person may have no social skills, no language skills, and major learning difficulties. At the other extreme the individual may have average or even above-average IQ and precocious vocabulary but odd social skills (e.g., being extremely self-centred). The former would receive a diagnosis of classic autism. The latter would receive a diagnosis of Asperger syndrome. Both of these are subgroups on the autistic spectrum, and those living with either condition share a strong preference for routines and repetition and are characterized by an obsessional interest in highly specific topics.

Prevalence. In the late 1990s the prevalence of autism spectrum conditions was found to have increased substantially relative to previous decades, and data published in 2009 revealed that the number of children diagnosed with these conditions was continuing to follow this same trend. A study published in December by the Autism and Developmental Disabilities Monitoring Network, a part of the U.S. Centers for Disease Control and Prevention, stated that in 2006 between 1 in 80 and 1 in 240 U.S. children had been diagnosed with an autistic condition. The study assessed the prevalence of these conditions in eight-year-olds, since this represented the age by which most affected U.S. children had been placed under a specialist's care. A similar prevalence estimate, about 1 in 100 children, had been reported previously in the United Kingdom. This figure was also representative of an increase in the number of persons diagnosed with autism spectrum conditions, relative to previous decades in the United Kingdom. It remained unclear, however, whether such increases were due to the conditions' becoming more widespread or to improvements in knowledge and diagnostic procedures used to detect them.

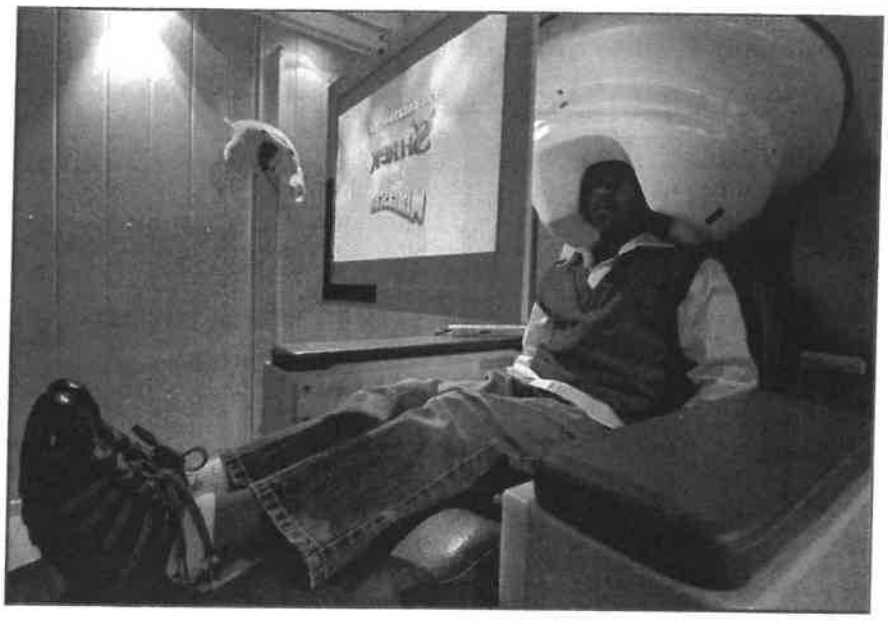
Information on the incidence of autism spectrum disorders in other countries remained controversial. A study published early in 2009 in the *Israel Medical Association Journal* addressed the incidence of autism spectrum disorders in that country. The study, which represented the first comprehensive analysis of autistic disorders performed in Israel, reported that the incidence of the conditions had increased from 2 diagnosed cases in 1985 to 428 in 2004. These figures were further assessed in terms of the total number of children under age 18 who lived in the country during the time period investigated. Thus, the per capita prevalence of autism spectrum disorders in Israel in 2004 was estimated to have been 1.9 per 10,000 children. A group of researchers from the Sheba Medical Center near Tel Aviv later wrote to the journal claiming that the reported figures were underestimates. The group stated that for children born

Drew Perine—The News Tribune/AP

in Israel between 2001 and 2004, the prevalence of autism spectrum disorders was about 36 per 10,000, placing the incidence of the conditions in the country closer to estimates of worldwide incidence reported in a study published in 2006 in the journal *Lancet*. The Israel figure cited by the group at Sheba Medical Center had not yet been published in a peer-reviewed journal and was based on data that the researchers had obtained from the Israel Ministry of Social Affairs.

Psychological Aspects. One of the most distinguishing characteristics of people affected by autism spectrum conditions is atypical behaviour. Such behaviour has been associated with two primary psychological aspects, empathy and systemizing. Empathy involves imagining another person's thoughts and feelings and having an appropriate emotional reaction to those feelings. Children and adults with Asperger syndrome show their empathizing difficulties on tests of emotion recognition, theory of mind, and spontaneous empathy. Theory of mind is the ability to attribute mental states to oneself or others and is regarded as the cognitive component of empathy. Emotion recognition is sometimes regarded as part of theory of mind because emotions are mental states. Often emotion-recognition deficits appear only if complex emotions are tested, though in some individuals with autism, the deficit is evident even when basic emotions are tested. This deficit can explain the difficulties in social and communicative development and in imagining others' minds.

Systemizing is the drive to analyze a system in terms of underlying rules in order to understand and predict its behaviour. People with autism spectrum conditions show precocious understanding of systems, relative to their mental age, on tests of intuitive physics or questionnaires assessing how interested a person is in different types of systems (maps, train timetables, machines, syntax, etc.). The repetitive behaviour, the desire for routines, and the need for sameness can be interpreted as being the result of a strong drive to systemize. Systemizing also requires excellent attention to detail, and individuals with autism or Asperger syndrome are relatively fast on tests of attention to detail. Adept systemizing can often explain the exceptional mental abilities possessed by some persons affected by these conditions.



An autistic child views a movie during a demonstration of magnetoencephalography (MEG). This technique can be combined with magnetic resonance imaging (MRI) to obtain information about abnormalities in brain function in individuals with autism.

Neurological Aspects. Anatomical abnormalities have been identified in different brain regions in individuals with autism. These abnormalities are not found in every case, and there are inconsistencies between studies, such that sometimes overgrowth or underdevelopment is found. The brain regions that have been reported to be atypical include the cerebellum, the corpus callosum, the hippocampus, and the amygdala. Epilepsy also occurs in a proportion of individuals with autism spectrum conditions, though the exact rate is not clear. Although in classic autism one-third of cases develop epilepsy by adolescence, in the Asperger subgroup these rates may be much lower and have not been systematically studied. In terms of neuropathology, the number of Purkinje cells (large neurons with many branching extensions) in the cerebellar cortex in people with autism is abnormally low. Abnormalities have also been reported in the density of neurons in the hippocampus, the amygdala, and other parts of the limbic system. One report also suggested a reduction in the size of cortical minicolumns, though the significance of this is unclear.

Functional neuroimaging studies of autism spectrum conditions show abnormalities in the amygdala and in the orbitofrontal and medial prefrontal cortex, among other areas. These atypical patterns of neural activity occur in relation to the empathizing deficits. Using magnetic resonance imaging (MRI), some reports have suggested that the autistic brain involves transient postnatal macroencephaly (accelerated brain growth). For example, in a study of children diagnosed with autism, some 90% who were born with normal head circumference were found to have, by ages two to four, MRI-based brain volumes that were larger than average. Independent confirmation of these abnormal growth rates is needed.

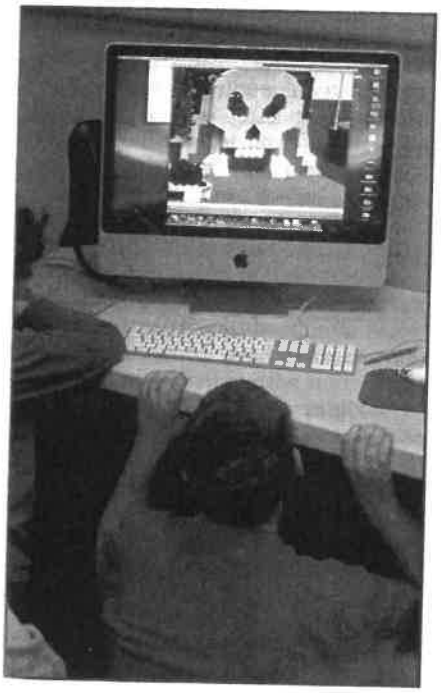
Genetic and Hormonal Aspects. In recent years the sibling risk rate for autism has been estimated to be about 5 to 10 times higher than general population rates. That is, while 1% of children have an autism spectrum condition, the sibling recurrence rate is 5–10%. Regarding twin studies where one twin had autism, 60% of MZ (identical) pairs were found to be concordant for autism, meaning that both twins were affected. In contrast, no pairs of DZ (fraternal) twins demonstrated concordance. Genetic linkage studies have led to the implication of a number of chromosomal regions in autism. In addition, abnormalities affecting the X chromosome have been identified in association with autism, which may explain the sex ratio's being markedly biased toward males. The marked sex ratio in autism may also reflect hormonal factors. For example, studies have indicated that levels of fetal testosterone are associated with the number of autistic traits present in an individual. Within typical development, fetal testosterone is negatively correlated with the later development of eye contact, vocabulary, empathy, and social

Jacek Kaczmarek/AP

skills. The hormone also is positively correlated with the emergence of narrow interests, systemizing, and autistic traits. Thus, the higher the levels of the hormone during fetal development, the more likely these features are to appear in early childhood.

Early Diagnosis. The earliest that classic autism has been reliably diagnosed is 18 months of age. This was shown by a screening approach known as the Checklist for Autism in Toddlers (CHAT), which tests for the absence of "joint attention behaviours," such as pointing and gaze following, and the absence of pretend play, all of which are typically present by this age. Population-based studies have shown that CHAT, developed by the U.K.-based Autism Research Centre, has excellent specificity—children who failed on this test had an 83.3% chance of developing autism or a related pervasive developmental disorder. The approach, however, has a low level of sensitivity—it detected only two out of every five cases, mostly missing the Asperger subgroup. Revisions of CHAT are under way to further improve the technique. Asperger syndrome was found to be reliably diagnosed by age five by using a screening technique called Childhood Autism Spectrum Test (CAST).

Autistic children gather around a computer to watch a movie made with Lego toys. Lego therapy has been shown to help children with autistic disorders develop their social communication and turn-taking skills.



In programs such as PEERS (Program for the Education and Enrichment of Relational Skills), at the University of California, Los Angeles, autistic teens develop important social skills through activities such as playing games with others.

Intervention. The most effective interventions for children on the autistic spectrum are special education, such as social skills teaching, and applied behaviour analysis (ABA), in which appropriate skills and behaviours are taught through principles of reinforcement. The key ingredients for effective early intervention are that the methods are highly structured, intensive, and individualized. Medical treatments are not usual. Indeed, there are ethical issues surrounding the notion of trying to cure autism. Although some aspects of the condition do require help (e.g., the empathy difficulties and the lack of language development), other aspects may not (e.g., the systemizing talents). For many years "treatment" in autism proceeded on the basis of an approach that was tried and tested but without any real rationale for why it should be effective. ABA is one such example. The principles behind it enable target skills to be broken down into simpler units to be acquired through shaping and mass practice. While there is some evidence for the effectiveness of ABA, the methods require external reinforcers or rewards to maintain the child's attention and cooperation, which suggests that they are not as autism-friendly as they could be. In contrast, newer interventions have been designed to harness individuals' areas of strength and their natural interests as a means for building new skills. One example is *Mind Reading* DVD educational software, in which the individual's natural interest in lawful, predictable computers and in information's being systematically organized renders the domain of emotions easier to learn about. A second example is *The Transporters* DVD animation, which relies on the child's natural interest in the mechanical, predictable motion of vehicles to help the child attend to the film and to enable implicit learning of emotions, since these are grafted onto the vehicles. A final example is Lego therapy, which also exploits the child's strong interest in systems (in this case, constructional systems) to encourage turn taking and social communication.

All treatments that are claimed to be useful for children or adults with autism spectrum conditions should be subjected to a scientific evaluation. This is to confirm that there are indeed benefits relative to no intervention and that there are no unwanted side effects. Web sites have been developed to help parents as well as professionals make informed decisions about which treatment option to pursue, given that these are often expensive and that new methods are announced almost annually.

Mental Health in Affected Teenagers and Adults. Teenagers and adults with Asperger syndrome often suffer from additional mental health problems, the most common being depression. Many also feel suicidal, and tragically some

(Bottom) Ron Tarver—MCT/Landov; (top) Kim Johnson Flodin/AP

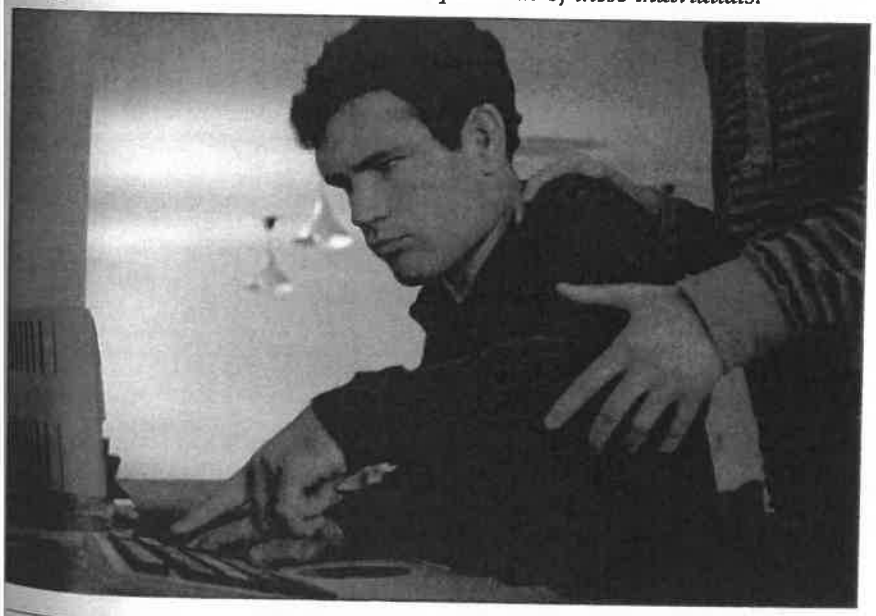
are so desperate as to attempt suicide. The high levels of depression are not surprising if people with Asperger syndrome feel that they do not fit into society and feel rejected by the majority. For teenagers this may be because the peer group becomes less tolerant of those who do not fit in and because a failure to conform in the educational system can also mean underperforming academically. In adulthood, depression may be associated with the challenges of living independently or with the difficulties in securing employment if the selection is via an interview (i.e., relying on social skills). Those adults with high-functioning autism or Asperger syndrome who are fortunate to have a job may find that they either lose their jobs (e.g., through interpersonal difficulties at work) or fail to be promoted (e.g., because they cannot manage others). Such depression may therefore be secondary to the condition and may be preventable with appropriate support. Such support for teenagers may include special education, a buddy system, social skills training, and organized social groups. Support in adulthood may include sheltered employment, mentoring, advocacy, befriending, social groups for Asperger syndrome, and help with housing.

The adult Asperger community has generated its own term for those who do not have Asperger syndrome or autism: *neurotypicals*. This is in part intended to convey that one view of autism spectrum conditions is that they are not a disease or a disorder but simply an atypical form of neurological development, akin to left-handedness. This view is less stigmatizing and makes an important political point—namely, that those individuals whose brains develop and work differently need not be judged to be inferior to the majority. They are simply "different." This view is called the neurodiversity movement and resembles the civil rights movement that was necessary to gain equal opportunities for ethnic minorities and for women. There is much to recommend this view, since the profile of autism spectrum conditions entails not just disability but also areas of strength (e.g., in attention to detail and systemizing). Nevertheless, it is important that the disability element is not overlooked in this shift of emphasis, since the diagnosis is given only if the individual is suffering to some extent, with their autistic traits leading to significant interference in their everyday life. This diagnostic criterion is important as a yardstick of severity, since if the features of the condition are very mild, they may not require a diagnosis. This relates back to the notion of an autistic spectrum, a dimension of autistic traits that runs right through the population. The current view is that all humans lie somewhere on this spectrum and that it is only those who are at the extreme end and those who are experiencing difficulties as a result of their high number of autistic traits who will require a diagnosis. Not all doctors share this view of autism spectrum conditions' simply being a case of neurodiversity. For example, the American Psychiatric Association retains the term *autism spectrum disorder,* since this group of conditions includes the pervasive developmental disorders, which involve varying degrees of impairment of language acquisition, communication, social behaviour, and motor function.

Those individuals on the autistic spectrum who also have below-average IQs (and therefore are at risk of broader developmental delays) are likely to need special support throughout their lives. They will remain vulnerable even as adults. The issues and challenges that low IQ raises are in some respects no different from those issues raised by low IQs in other individuals (who are not on the autistic spectrum) and are probably best served by the same learning disability agencies.

An autistic teenager holds his puppy and a small bucket of his belongings after receiving a Project Lifesaver tracking bracelet at the Staunton (Va.) Police Department.

Communities for autistic adults, such as Cascina Rossago in Italy, create a living environment that caters to the unique needs of these individuals.



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(Bottom) Silvia Morara/Corbis; (top) Mike Tripp—The Daily News Leader/AP