

Autism

SIMON BARON-COHEN

Introduction

Autism, a condition of neurodevelopment, is more common in males, with onset typically in infancy. It is diagnosed when a child or adult has abnormalities in a ‘triad’ of behavioral domains: social development, communication, and repetitive behavior / obsessive interests. People with autism typically may appear to be self-absorbed, or ‘in a different world,’ due to their lack of responsiveness to those around them. It can occur at any point on the IQ continuum, and IQ is a strong predictor of outcome. Autism is also invariably accompanied by language delay (e.g., no single words before 2 years old). Asperger syndrome (AS) is a sub-group on the autistic spectrum. Those with AS share many of the same features as are seen in autism, but with no history of language delay and their IQ is in the average range or above. Typically, classic autism is identified before age 3, and can be diagnosed as early as 18 months. AS is typically identified later than this, usually around age 6 years old.

Frequency and clustering in the population

Early estimates of the prevalence of autism suggested it was quite rare (4 in 10,000 children), but over the 1990s a series of reports suggested that it was far more common than this. The highest estimate is that autism spectrum conditions (i.e, autism and AS) occur at a rate of 1 child in every 166. This massive change in reported figures has been interpreted by some to indicate that there is some new environmental cause (e.g., vaccine damage), but critics of this view argue that the increase can be explained more simply as the result of widening the diagnostic category (e.g., previously AS was not included). Other explanations have included improved awareness among clinicians and the general public (previously there was no special training or media coverage of autism), and the existence of many services for autism (which hitherto simply did not exist).

Psychological causes of autism

It is widely recognized that people with autism spectrum conditions process information differently to other people. Information as a psychological construct is referred to as cognition. The main cognitive theories of autism are summarized below.

Mindblindness theory

The mindblindness theory proposed that in autism spectrum conditions there are deficits in the normal process of empathizing, relative to mental age (Baron-Cohen, 1995). These deficits can occur by degrees. The term ‘empathizing’ encompasses a range of other terms: ‘theory of mind,’ ‘mind-reading,’ and ‘empathy,’ and involves two major elements – (a) the ability to attribute mental states to oneself and others, as a natural way to make sense of other people; and (b) having an emotional reaction that is appropriate to the other person’s mental state. In this sense, it goes beyond what is normally meant by the term ‘theory of mind’ to include having some affective reaction (e.g., sympathy).

Since the first evaluation of mindblindness, there have been more than thirty experimental tests. The vast majority of these have revealed profound impairments in the development of empathizing ability in people with autism. This is manifested in the form of reduced shared attention, reduced use of mental state terms in language, reduced production and comprehension of pretense, difficulties in appreciating other people’s different beliefs, and difficulties in understanding subtle emotions.

Empathizing-Systemizing (E-S) theory

Systemizing is the drive to analyze and build systems, in order to understand and predict the behavior of events that do not involve agents. Systems are all around us in our environment, and include technical systems (e.g., machines and tools), natural systems (e.g., biological

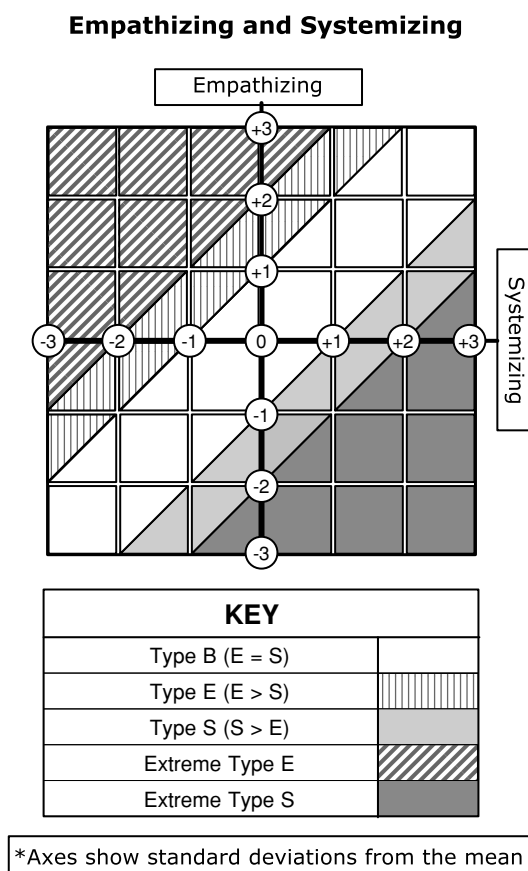


Figure 1. Empathizing and Systemizing. These two psychological processes are plotted as independent dimensions (E and S). Research on sex differences in the general population shows that more females than males have stronger E relative to S ($E > S$), and that more males than females have stronger S relative to E ($S > E$). People on the autistic spectrum tend to show intact or even superior S, alongside impaired E ($S \gg E$), relative to controls matched for mental age. Put succinctly, females are more likely to have a brain of Type E, males are more likely to have a brain of Type S, and people with autism are more likely to have a brain of Extreme Type S.

and geographical phenomena), abstract systems (e.g., mathematics or computer programs), and even social systems (e.g., profits and losses in a business, or a football league table). The way we make sense of any of these systems is in terms of underlying rules and regularities, or specifically an analysis of input-operation-output relationships.

E-S theory holds that alongside their deficits in the ability to empathize, the ability to systemize is either intact or superior in people with autism (Baron-Cohen, 2002). Initial studies of systemizing are consistent with these predictions. The systemizing talent found in a significant proportion of people with autism may help account for why 'islets of ability' in music, mathematics, drawing, and memorizing lists of information (e.g., train time-tables, calendrical calculation) may be found more often in autism than in other neurodevelopmental conditions. Because in the general population,

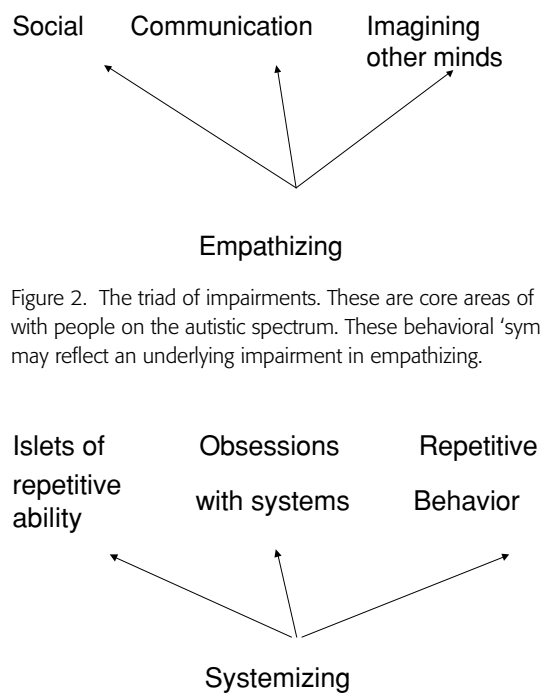


Figure 2. The triad of impairments. These are core areas of difficulty with people on the autistic spectrum. These behavioral 'symptoms' may reflect an underlying impairment in empathizing.

Figure 3. The triad of strengths. These are core qualities that are usually very strong in people with autism, and may not be unwanted. Islets of ability may be expressed as a talent in one skill, or at least an uneven cognitive profile relative to the person's other skills. Obsessions often focus on systems of one kind or another, and are an example of the person with autism going into a topic or activity in great depth, becoming a 'specialist.' Repetitive behavior was traditionally viewed as undesirable. These core qualities may, however, reflect an intact or even superior drive to systemize.

empathizing tends to be stronger in females and systemizing tends to be stronger in males, it has been proposed that autism may represent an 'extreme of the male brain.'

Executive dysfunction theory

People with autism spectrum conditions show 'repetitive behavior,' a strong desire for routines, and a 'need for sameness.' To date, the only cognitive account to attempt to explain this aspect of the syndrome is the executive dysfunction theory (Ozonoff *et al.*, 1994; Pennington *et al.*, 1997; Russall, 1997). This paints an essentially negative view of repetitive behavior, assuming that it is a form of frontal lobe perseveration (a tendency to repeat the response to a particular situation in other, inappropriate conditions), or an inability to shift attention. People with autism who have additional learning disabilities are more likely to show executive deficits. But the fact that it is possible for those with AS to exist who have no demonstrable executive dysfunction, while still having deficits in empathizing and talents in systemizing, suggests that executive

400 Developmental pathology

dysfunction cannot be a core feature of autism spectrum conditions.

The executive account has also traditionally ignored the content of repetitive behavior. The E-S theory, in contrast, draws attention to the fact that much repetitive behavior involves the child’s ‘obsession’ with, or strong interest in, mechanical systems (e.g., light switches, water faucets), or other systems that can be understood in terms of rules and regularities. Rather than these behaviors being a sign of executive dysfunction, they may reflect the child’s intact or even superior interest in systems. A study of obsessions suggests that autistic obsessions are not random with respect to content, which would be predicted by the content-free executive dysfunction theory, but that these tend to cluster in the domain of systemizing.

Central coherence (CC) theory

‘Weak’ central coherence refers to the individual’s preference for local detail over global processing (Frith, 1989). People with autism have been demonstrated to have a superior ability in the Embedded Figures Task – a test that involves locating a hidden part within a whole design as quickly and accurately as possible. This confirms that in autism there is superior attention to detail. Systemizing requires, as a first stage, excellent attention to detail, identifying parameters that may then be tested for their role in the behavior of the system under examination. So both the E-S theory and the CC theory predict excellent attention to detail. However, both theories also make opposite predictions when it comes to an individual with autism being able to understand a whole system.

The E-S theory predicts that a person with autism, faced with a new system to learn, will show a stronger drive to learn the system, compared to someone without autism, as long as there are underlying rules and regularities that can be discovered. Moreover, they will readily grasp that a change of one parameter in one part of the system may have distant effects on another part of the system. In contrast, the CC theory should predict that they will fail to understand whole (global) systems or the relationships between parts of a system. This prediction has not yet been tested.

Neurological causes of autism

A neural basis of empathy or social intelligence was first proposed by Brothers (1990). She suggested, from animal lesion studies, single cell recording studies, and neurological studies that social intelligence was a function of three regions: the amygdala, the orbitofrontal cortex (OFC), and the superior temporal

sulcus and gyrus (STG). Together, she called these the ‘social brain.’ There are several important lines of evidence implicating the amygdala in primate social behavior.

Amygdala-lesioned monkeys become socially isolated. They fail to initiate social interactions, and to respond appropriately to social gestures. When the amygdala-lesioned monkeys were released into the wild, they were unresponsive to group members, failed to display appropriate social signals (both affiliative and aggressive), and they withdrew from other animals.

Jocelyne Bachevalier lesioned either the medial temporal lobe (including the amygdala), or just the amygdala. The lesioned animal infants were raised and paired with an age-matched control animal. At 2 months, the infants with medial temporal lobe lesions were more passive, displayed increased temper tantrums, and initiated fewer social contacts. At 6 months, they interacted very little with the control animal, and actively withdrew from all approaches by the normal animals. Those with medial temporal lobe lesions also displayed emotionally expressionless faces, and showed more self-directed behavior and stereotypies (repetitive movements). Such abnormalities were still evident in adulthood. Amygdala lesions alone produced a similar pattern of social abnormalities, but to a lesser extent.

The human amygdala is activated in humans when decoding signals of social importance, such as gaze, expression-recognition (especially of fearful faces), and body movements. There are several lines of evidence for an amygdala deficit in autism. A neuroanatomical study of autism at post-mortem found increased cell density in the amygdala, although its volume was normal. In addition, patients with amygdala lesions show impairments in social judgment that have been likened to ‘acquired autism.’ The age of onset of deficits in acquired autism compared to idiopathic cases is likely to mean that the two syndromes also differ in many ways. Likewise, patients with autism tend to show a similar pattern of deficits to those seen in patients with amygdala lesions. A recent structural magnetic resonance imaging (sMRI) study of autism reported reduced amygdala volume, while in a recent functional magnetic resonance imaging (fMRI) study, adults with autism but with normal intelligence, or with AS, showed significantly less amygdala activation during an empathizing task.

While these sorts of findings highlight the likely role an amygdala abnormality might play in autism, it is likely that it is not the only abnormal neural region. For example, the case has been made for anomalous functioning in the cerebellum, hippocampal formation, medial frontal cortex, and fronto-limbic connections in autism. Reduced neuron size and increased cell-packing

density has also been found in the limbic system, specifically the hippocampus, subiculum, entorhinal cortex, amygdala, mammillary bodies, anterior cingulate, and septum in people with autism. Research is also focusing on the role of neurotransmitters, especially serotonin, in the autistic brain.

Genetic causes of autism

Ultimately, the behavioral, cognitive, affective, and neural abnormalities in autism spectrum conditions are likely to be due to genetic factors, as there is evidence for autism and AS being strongly heritable. Moreover, studies in molecular genetics are beginning to narrow down candidate regions on chromosomes (e.g., chromosome 7) that might be different in autism (International Molecular Genetics Study of Autism Consortium [IMGSAC], 2001).

Conclusions

Currently, there are no specific medications that are recommended for autism, but it is recognized that early identification of children with autism spectrum conditions is desirable, and special education in the form of highly structured, intensive programs is beneficial. Some of these programs are based on techniques from speech therapy (including the encouragement of social communication, such as joint attention behavior), whilst others are reward-based behavioral programs, where the children can use their natural systemizing skills to learn

rules for social interaction. Some new tailor-made educational software is also being developed for people with autism, capitalizing on the fact that many of them enjoy using computers to learn about the world. The future of research in this field will be to understand the relationships between these different causal levels in autism, and to evaluate the most effective treatments.

See also:
Theories of the child’s mind; Magnetic Resonance Imaging; Brain and behavioral development (I): sub-cortical; Brain and behavioral development (II): cortical; Executive functions; Face recognition; Intelligence; Selfhood; Socialization; Behavior and learning disorders; Behavior genetics; Cognitive neuroscience; Ethology

Further reading

Baron-Cohen, S. (2003). *The Essential Difference: Men, Women and the Extreme Male Brain*. London: Allen Lane.
Baron-Cohen, S. and Bolton, P. (1993). *Autism: The Facts*. Oxford: Oxford University Press.
Wing, L. (2002). *The Autistic Spectrum*. London: Constable Robinson.

Acknowledgments

The author was supported by the MRC during the period of this work. Portions of this article also appeared in *Encarta* (2004).