Autism

Autism is a spectrum condition, in that it manifests in varying degrees of severity. At one extreme, a person may have no social skills, no language, and major learning difficulties. At the other end of this spectrum, a person may have normal or even above average IQ, precocious vocabulary development (though a lack of interest in small-talk or chatting), and social skills that are only odd by virtue of being one-sided or extremely self-centred. The former case would receive a diagnosis of classic autism. The latter case would receive a diagnosis of Asperger Syndrome (AS). Both represent subgroups on the autistic spectrum. Both also share a strong preference for routines and repetition, and where the intellectual style narrow and deep – an 'obsessional' interest in highly specific topics. Up to 1% of the population are somewhere on the autistic spectrum.

Psychological aspects

The empathising-systemising (E-S) theory[1] proposes that there are empathising deficits in autism, whilst systemising is either intact or superior. Empathy involves imagining another person's thoughts and feelings and having an emotional reaction that is appropriate to that other person's feelings. Children and adults with AS show their empathising deficits on age-appropriate tests.[2] This deficit underlies the difficulties in social and communicative development and in imagining others' minds. Systemising is the drive to analyse 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.[3] The unusually strong repetitive behaviour, the strong desire for routines, and the 'need for sameness', can be seen as the result of the person's strong drive to systemise. Systemising also requires excellent attention to detail, and people with autism and AS are faster on visual search tasks.[4] The strong systemising underlies the strengths that people with autism and AS have.

Neurobiological aspects

Anatomical abnormalities have been identified in many brain areas in autism. These include the cerebellum, corpus callosum, hippocampus, and the amygdala. Epilepsy also occurs in classic autism. In terms of neuropathology, the number of Purkinje cells in the cerebellar cortex is abnormally low.[5] Abnormalities in the density of packing of neurons in the hippocampus, amygdala, and other parts of the limbic system have also been reported.[6]

Functional neuroimaging suggests increased activity in sensory areas of the brain normally associated with stimulus-driven processing, and decreased activity in areas normally associated with higher-cognitive processing.[7] Abnormalities in autism have also been found using functional neuroimaging in the amygdala, the orbito and medial frontal cortex.[8] These atypical pattens of neural activity are associated with the empathising deficits. Using either MRI volumetric analysis, or measures of head circumference, the autistic brain appears to involve transient postnatal macroencephaly.[9]

Genetic and hormonal aspects

The sibling risk-rate for autism is approximately 4.5%, or a four-fold increase over general population rates. Regarding twin studies, when a narrow phenotype is considered, 60% of MZ pairs are concordant for autism vs. no DZ pairs. When a broader phenotype is considered, 92% of MZ pairs are concordant vs. 10% of DZ pairs.[10] Molecular linkage genetic studies have led to several chromosomal regions being identified, such as 2q, 7q, and 15q (22-24). Loci on the X chromosome have also been implicated in autism, which may explain the sex ratio in autism (markedly biased towards males).[11] The marked sex ratio in autism may also reflect hormonal factors. Currently there are clues that foetal testosterone (FT) may play a role: within normal development, FT is inversely correlated with frequency of eye contact, rate of vocabulary development, empathy and social skills, and FT is positively correlated with narrow interests and systemising.[12]

Early diagnosis and intervention

The earliest age at which classic autism has been reliably diagnosed is at 18 months of age, following a screening using an instrument called the CHAT (Checklist for Autism in Toddlers) which tests for the absence of 'joint attention' behaviours such as pointing and gaze following, and the absence of pretend play, all behaviours that are normally present by this age.[13] Asperger Syndrome has been reliably diagnosed by age five years, following a screening using an instrument called the CAST (Childhood Asperger Screening Test).[14] The most effective interventions for children with autism or AS are special education, such as social skills teaching, and Applied Behavioural Analysis (ABA), where appropriate skills and behaviours are taught through principles of reinforcement.[15] The key ingredients for effective early intervention are that the methods are highly structured, intensive, and individualised. Appropriate cognitive interventions are also beneficial for teenagers and adults. Medical treatments are not usual, and there are ethical issues surrounding the notion of trying to 'cure' autism, since whilst some aspects of the condition do require help (e.g. the empathy difficulties), other aspects may not (e.g. the systemising talents).

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Resources

- The National Autistic Society is the main charity in the UK for families with a child on the autistic spectrum: www.nas.org
- The Autism Research Centre, Cambridge University, contains a searchable database of publications and screening instruments such as the CAST, AQ, and CHAT: www.autismresearchcentre.com
- As interventions are scientifically evaluated, the results of such studies are summarised at www.researchautism.net

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