

The cognitive basis of a biological disorder: autism

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This article summarizes recent evidence indicating that individuals suffering from autism have a specific problem in understanding intentions and beliefs. We propose that this problem arises because they are incapable of forming a special kind of mental representation. A single cognitive deficit defines what is common to all autistic individuals. In contrast there is a wide range of proposals for the biological origins of the disorder.

Autism is a severe developmental disorder affecting an estimated 80000 individuals in Britain alone. The incidence of the most classical form of autism is 2-5 in 10000 births, and there are three times as many males with autism as females²; almost all need special education and lifelong support³. The condition of autism was first identified in 1943 by Kanner⁴. Then, and now, autism is defined behaviourally and so far the earliest confident diagnosis is at two-and-a-half years⁵. Because of current diagnostic uncertainties, many autistic individuals never receive an official diagnosis. It is now increasingly thought that there may be a spectrum of autistic disorders from mild to severe, including different variants of autism⁶.

All variants have three core features in common⁷: (1) impairment in socialization (a specific impairment in the quality of reciprocal interactions); (2) impairment in communication (a delay in language acquisition and poor use of verbal and non-verbal means of communication); and (3) impairment in imagination (a lack of spontaneous pretend play). These impairments do not co-occur by chance⁸. If these features are used for diagnosis, the incidence of autistic spectrum disorders may be as high as 2-3 per 1000 births⁹.

Many of the striking features that can be observed in individuals with autism at various ages are now being recognized as secondary social consequences of autism. Such symptoms include social avoidance and anxiety¹⁰. A number of cognitive deficits have previously been proposed as central to autism; these include problems of object permanence¹¹, language¹² and perception¹³. When these have been investigated as potentially causal problems of autism, they have all been found insufficient or irrelevant^{14,15}. After taking into account developmental level [mental age (MA)], degree of mental retardation (IQ) and chronological age (CA), many supposedly typical symptoms were found to be neither unique nor universal to autism¹⁶.

It is now generally accepted that there must be a biological origin to autism¹⁷. There are several reasons for this. First, it is known that people with autism have a greatly increased chance of having diverse medical conditions as background factors¹⁸. Certain conditions occur more often along with autism than others. In a total population study of Scandinavians with autism (n = 35), epilepsy

affected one third of cases by early adulthood, major EEG abnormality affected ~45% of cases, severe perinatal distress affected ~28%, neurogenic hearing deficit affected ~20%, while 4% of cases in this study also showed major CSF-protein abnormalities. Second, 37% of cases in this population displayed known medical syndromes such as Fragile X, tuberous sclerosis, neurofibromatosis, Rett syndrome and congenital hydrocephalus¹⁸. Third, autism is strongly associated with mental retardation¹⁹. As progressively more retarded samples of children are taken, the likelihood of autism increases²⁰. We know that the retardation does not of itself cause autism; many Down's syndrome children, in particular, show a high degree of sociability²¹. Furthermore, in a quarter of cases, we find autism without retardation.

One simple model for this pattern is that there is a specific brain system necessary for normal development, the disturbance of which leads to autism²². One possible candidate is the dopamine system²³. Brain damage that results in general intellectual retardation could be seen as due to randomly distributed lesions. The more the damage, the higher the probability that this particular brain system will be affected. In addition, there is the possibility of direct damage to the critical system alone.

Even in people with relatively mild degrees of autism, magnetic resonance imaging (MRI) techniques have revealed abnormalities in the cerebellar vermis²⁴. Autopsy studies have suggested neuronal disruption in a number of brain areas, particularly in the limbic system²⁵, and recent reviews of the literature conclude that there is cortical as well as subcortical involvement²⁶. However, such studies have not yet separated causal and correlative relationships between brain damage and autism. As to the biological origins of autism, genetic factors have recently received particularly strong confirmation²⁷. However, other causal factors, such as perinatal problems, immunological deficiency and viral disease are being considered as well²⁸.

It is clear that any theoretical account of autism must include its biological basis, yet the gulf between the brain and complex behaviour is great. The causal chain between candidate biological factors and the resulting behavioural impairment requires an intervening cognitive level^{22,29}, as shown in Fig. 1 where a distinction between four different levels of description is drawn. Note that we use the term 'cognitive' in its current broadest sense, which would include, not exclude, affective factors. While facts can be collected directly at the biological and behavioural level, inferences have to be drawn about the cognitive level.

Given that autism has no single biological base, what justifies the application of a single label? We

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FACTS	<p>High genetic risk</p> <p>High incidence of associated medical disorders</p> <p>High incidence of mental retardation, neurological abnormalities, reflex abnormalities, seizures.</p>	Biological
QUESTION	<p>??? WHAT UNDERLIES ???</p> <p> THE BEHAVIOURAL SYMPTOMS</p>	Cognitive
FACTS	<p>Manifestation of triad of impairments varies with age and ability: core symptoms in areas of socialization, communication and imagination.</p>	Behavioural
FACTS	<p>Interpersonal problems: problems in family and peer group relationships, no friendships, no marriage, emotional repercussions, social avoidance.</p>	Social

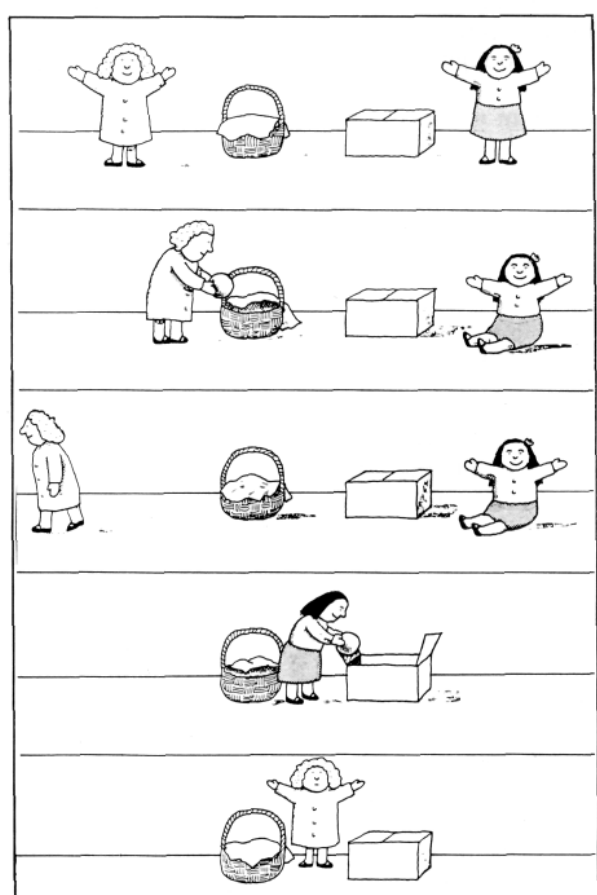
ascribe it to someone else; for example, in a form such as 'Some monks believed that "ducks are fish".' Second-order representations - in this case the representation of someone's belief — can be used to predict people's behaviour. For instance, monks could eat ducks on Fridays if they believed them to be fish. In this way we can establish relationships between external states of affairs and internal states of mind. We suggest that awareness of mental states and an emerging theory of mind are the result of a specific mechanism, which relates together yet keeps separate first- and second-order representations. This mechanism makes available a set of computational functions and these subserve different mental attitudes: 'pretend' for imagination, 'inform' and 'ask' for

Fig. 1. Levels of description relevant to autism.

propose that what all people with autism have in common is a particular cognitive deficit that gives rise to the core symptoms in the course of development³⁰. Before we can define this deficit and show how it accounts for the core symptoms, we have to draw attention to a particular aspect of normal development that has been explored only recently³¹, and that appears to be missing in autism. This is the development of the 'theory of mind'³², or 'mentalizing' - our ability to predict and explain the behaviour of other humans in terms of their mental states³³. Our ability to mentalize is revealed in our use and understanding of such words as 'believe', 'know', 'wish', 'desire', 'intend', and 'pretend'. A central feature of our proposal is that children with autism lack this ability.

The ability to mentalize is dependent on a specific mechanism that does not manifest itself from birth; neither can it be explained by learning³⁴. By age one, infants already attend to behaviour and internally represent many physical states of the world³⁵; i.e. they can remember and manipulate in their heads what they perceive in the world. These are first-order representations. From some time in their second year, children have at their disposal second-order representations and can in principle represent mental states as well as physical states. From this time the child not only attends to the behaviour of others, but has the means to learn to make sense of this behaviour by deducing the underlying mental state. Thus, by about age four, the normally developing child will have acquired a good understanding of the concept of 'belief'; i.e. that people can have different beliefs, including false beliefs.

What is the difference between first- and second-order representations? We know that 'ducks are fowl' and can represent that idea in memory as a first-order representation. At the same time, we can represent the idea 'ducks are fish' as long as we



Box 1. The basic test was in the form of a puppet show. Sally, one of the puppets, has a marble. She puts the marble in a basket and then leaves the room. Anne, the other puppet, takes the marble out of the basket and puts it in a box. She leaves. Sally comes back. She wants to play with her marble. Where will she look? See text for the results of this experiment. (Figure kindly supplied by A. Scheffler.)



Box 2. Another example of a false-belief attribution task. The typical child with autism, like the normal three-year-old, behaves as we have illustrated – as if other people would know what they themselves know. (Figure kindly supplied by A. Scheffler.)

communication, and 'believe' and 'intend' for socialization.

Let us take 'pretend' as an example. A two-year-old boy watches his mother speaking into a banana as if it were a telephone and he understands and remembers this game the following day. The memory 'Mother pretends "the banana is a telephone"' is a second-order representation and is different from an earlier memory 'the banana is good to eat'. The latter is an example of a first-order representation - something that happens to be true in the real world. One difference between first-order and second-order representations lies in the inferences that will be drawn. If you know that 'bananas are fruit' and you also learn that 'oranges are fruit', then your mental machinery will automatically work out that 'oranges are good to eat' but would not conclude that 'oranges are telephones'. The reason is that second-order representations are treated as exempt from confirmation from other knowledge or from the world, and by the same token are not used to generate new 'real world' inferences.

Pretend play contrasts with functional play, in which, for example, a miniature copy of an object is ascribed the properties of that object. If a child plays with a toy saucepan on a toy stove, making the kinds of movement, such as stirring and shaking, that mother makes on the real stove, this may be

merely functional play elicited by the physical properties of the toy. If the child sniffs and says that there is bacon frying and varies the play according to the dish, then this is pretend.

Precisely this type of play is hard to find in autistic children^{36,37}. By postulating that the cognitive cause of autism is damage to the 'theory of mind' mechanism, we could explain difficulties with the pretend function but, more importantly, we could also predict special problems with such computational functions as 'inform' and 'ask' and 'believe' and 'intend'. The first prediction concerning our theory was that, taking into account CA and MA, children with autism will have special problems in understanding belief.

The experiment illustrated in Box 1 checked this prediction³⁰. We have no problem in knowing that Sally will look for her toy in its hiding place in spite of our knowing that it is not there. This means that we can represent Sally's false belief as well as represent the true state of things. Normal children have no problems with this sort of task from about four years of age. Down's syndrome children with a mental age of five or six can also answer correctly. However, of a group of 20 autistic children, with a mean mental age of nine years, 16 failed the task in spite of being able to answer correctly a variety of



Box 3. Test of the 'inform' and 'ask' function. The normal child supplies the missing information; the child with autism often repeats what is already known. (Figure kindly supplied by A. Scheffler.)

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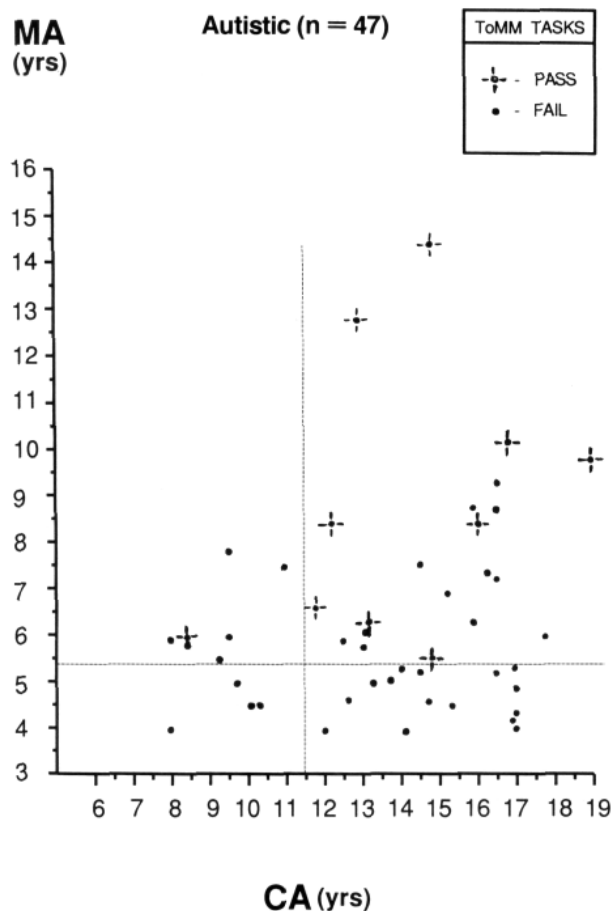


Fig. 2. Successful performance on mentalizing tasks as a function of mental age (MA) and chronological age (CA) in a sample of 47 children with autism. CA ranged from 8 to 19 years and verbal MA (using the British Picture Vocabulary Scales) ranged from 4 to 14 years.

questions of fact about what happened. They knew where Sally had put the marble, they knew that it was Anne who had moved it and that Sally had not seen the move. Their problem did not lie in perception, in memory or in language. The children with autism could not conceptualize the possibility that Sally believed something that was not true.

A number of experimenters have now carried out similar studies worldwide, confirming the specific impairment that autistic children have with beliefs³⁸⁻⁴⁸. Box 2 illustrates another variant of the test. Specific difficulties with the function 'inform' and 'ask' were also found (Box 3)⁴⁴.

In all these studies there was a minority of autistic children who performed the mentalizing tasks correctly. We ourselves have now tested 47 able children with autism on the false-belief and related tasks. The results from this study are shown in Fig. 2. With only one exception, no child with autism succeeded on a mentalizing task unless they were over eleven-and-a-half years old with a mental age of at least five and a half. In fact, most of those who succeeded had a mental age well over eight. As can be seen in the figure, even this high ability and age does not guarantee success. Could the success-

ful children have acquired some mentalizing ability after all, perhaps after a long delay? This explanation seems unlikely, since they still fail another, slightly more complex problem of the same kind ('John thinks that Mary thinks that. . .'⁴⁹), which children with Down's syndrome can solve and which should have been well within the intellectual capacity of the children with autism⁵⁰. Our preferred explanation at present is that these children have learned enough of human reactions to find their way round a restricted set of problems. It is as if they have extracted rules such as: 'When something in the world changes, people who just happen not to have seen the change occur behave (for some reason) as if they do not know about these changes.' Normal children do not acquire their mentalizing skills in this way⁵¹. However, as our analysis suggests, even those autistic children who solve the false-belief tasks find normal actions and reactions often totally irrational. This explains their continued problems with fluid mentalizing in everyday situations. One able man with autism complained that 'other people seem to have a special sense by which they can read other people's thoughts'¹⁴.

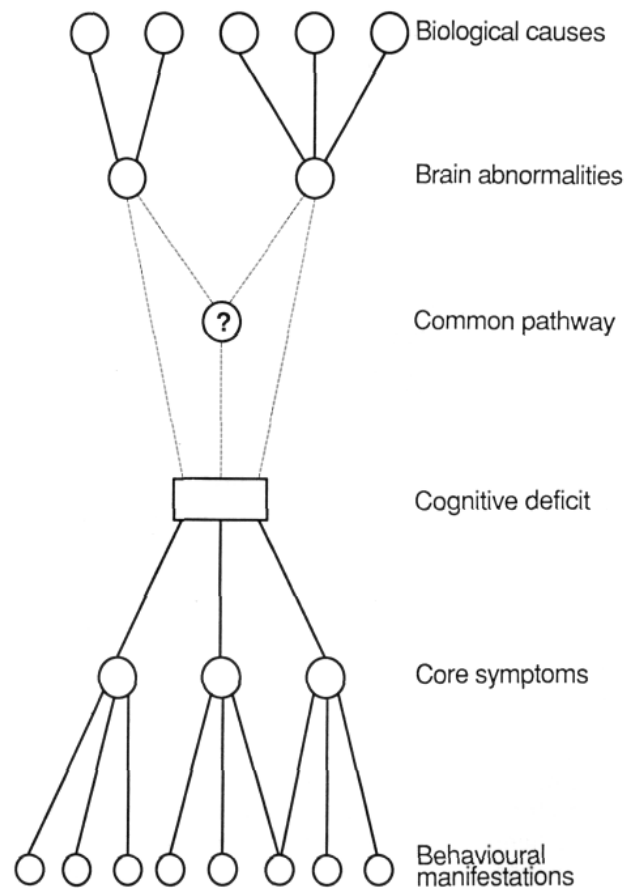


Fig. 3. A schematic representation of the causal analysis of autism. Such diagrams can be used for any neuro-physiological disorder.

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As illustrated in Fig. 3, we propose that the whole symptom complex of autism arises from a single cognitive deficit. This means we eventually need to derive from this single origin various types of behaviour, all associated with the core symptoms but changing with age and ability. We believe this can be done, although some symptoms — notably those associated with repetitive actions and special skills — may need additional explanation¹⁵. One should note that there are alternative formulations of the cognitive hypothesis⁵²⁻⁵⁵: some of these stress the importance of early affective factors in inter-personal relationships that may be missing from birth in autism. However, evidence on this point remains to be obtained. Our present account suggests that if autistic children have a faulty mentalizing ability and cannot form second-order representations, then they will not create a normal 'theory of mind' and will not understand that people can have beliefs, feelings and other attitudes. Without such knowledge, social relationships will be difficult to form and maintain and empathy will be almost impossible.

In terms of social relations, children with autism are sometimes described as treating objects and people alike, although, in fact, their behaviour can vary from complete withdrawal (the most well-known symptom) to repetitive pestering of other people⁵⁶. Both extremes can be explained by a lack of 'theory of mind'. Clearly, if autistic individuals do not realize the effect of their behaviour on other people's attitudes, and if they have no awareness of others and self as mentalizing beings, then their social interactions will be peculiar.

Communication problems in autism arise both verbally and non-verbally and encompass a wide range of difficulties⁵⁷⁻⁵⁸. While phonological development is appropriate for mental age, there is usually a delay in other aspects of language acquisition. Autistic individuals have particular problems with the pragmatic aspects of language and have difficulties in handling mental state expressions⁵⁹. Even those who eventually acquire fluent language are very literal, be it in terms of comprehending metaphor or in terms of understanding the indirect utterances we use normally in conversation. Thus, if asked 'Can you pass the salt?' a child with autism is prone simply to answer 'yes', rather than treating the question as a request. Such behaviour does not arise due to a lack of intelligence or a misplaced sense of humour; it comes from an inability to understand that there are intentions behind words.

The relationship between biology and cognition

We have attempted to show how the cognitive account explains the three core symptoms of autism. What, then, is the relationship between the cognitive deficit and the possible biological causes? The question itself reveals that autism is a disorder that has a (necessary and sufficient) causal definition at the cognitive level but not at the biological level. Thus, what we want to find is some biological system, equivalent to the fully functioning 'theory of mind' mechanism, the development of which can be

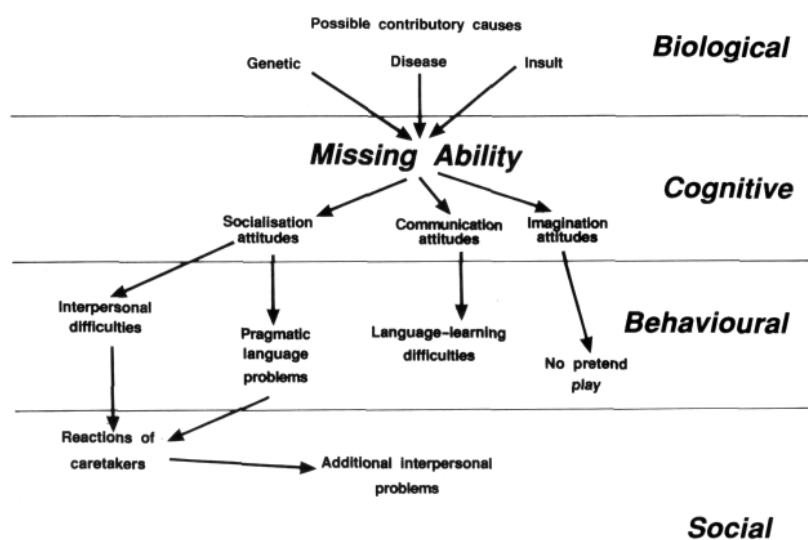


Fig. 4. A multi-level causal analysis of the autism syndrome, showing our proposals for the relationships between the various behavioural manifestations and their cognitive origins. The diagram also indicates how biological factors start the causal chain and how social consequences are derived from the behaviour. Feedback from social to cognitive and behavioural levels has been omitted.

disrupted in a variety of ways. The dopamine system is an interesting example that could fill such a role. We suggest that the multiplicity of biological factors will lead to a limited set of disruptions, which in turn will lead to failure of the theory of mind mechanism. These factors might include, for example, developmental malformation of the structure underlying the mechanism or disconnection of the mechanism.

These relationships are revealed in Fig. 4. Note that we are not elevating any symptom to causal status. The cause we cite — the 'missing ability' in the diagram — we have hypothesized to be a particular computational deficit to do with the creation of second-order representations^{34, 54}. If we are wrong, and the missing ability turns out to be some other cognitive function (again, using 'cognitive' in its broadest sense) Fig. 4 would scarcely be affected. On the other hand, if we ignore the cognitive aspects of our causal theory, we would then have to build a whole series of separate causal bridges between the biological base and the behaviour.

In Fig. 3 we have presented a schematic version of the causal diagram. This helps us to appreciate the value of the method independently of the particular hypothesis we currently hold. The same causal reasoning would apply if an attentional or affective hypothesis or any other non-social hypothesis of the origins of the disorder was adopted. Of course, it would be incumbent on the proposer of any hypothesis to establish a satisfactory causal nexus covering the full range of behavioural symptoms. Figure 3 suggests that a convergence in the causal nexus of autism can be located at the cognitive

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level. If future research comes to the conclusion that the core impairments in autism are different and have different underlying causes, then this convergence would vanish, and, in the absence of a convergence at the biological level, the justification for the single term 'autism' would be removed.

The option to define autism in biological terms carries with it certain implications. Either a convergence must be demonstrated from a faulty gene, a fragile X chromosome, maternal rubella, a viral illness and all the other possible causal origins onto a single biological node that will have a fixed description in relation to the cognitive deficit (e.g. a specific disruption in the maturation of the structures underlying the theory of mind mechanism), or we have to define subtypes of autism for all the biological manifestations. If there was no common pathway in the illustration in Fig. 3, there would be two such subtypes. Indeed, subclassifications can be imagined at all the levels we have discussed as being valid for different purposes.

In development, neural systems interact with information from the environment to form more complex neural systems, which have specific functions. In certain parts of the brain, these functions are cognitive functions, and the underlying neural systems are also cognitive systems. Cognitive systems might be localized or distributed in the brain. Some biological abnormalities can cause cognitive abnormalities during development. The intellectual enterprise that unites biologists and psychologists is to trace this relationship. Causal analyses are an essential part of this endeavour.

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