Autism Spectrum Disorder

Characteristics, Causes and Practical Issues

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A FRAMEWORK FOR EXPLAINING AUTISM

WHY EXPLAINING AUTISM IS IMPORTANT
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SUMMARY
What Causes Autism?

**AIMS**

The main aim of this chapter is to establish a framework for thinking about causal explanations of autism, to be used in subsequent chapters in Part II. Additional aims are to stress the importance of understanding the causes of autism, and to foster a critical approach to theoretical explanations of autism.

**WHY EXPLAINING AUTISM IS IMPORTANT**

It is of considerable practical importance to understand the causes of autism. Better understanding is needed to reduce the prevalence and symptom severity of debilitating forms of ASD that are associated with low learning and language ability and multiple medical and behavioural problems. The desirability of preventing or curing ‘pure’ autism in high-functioning individuals is questionable (see the opening section of Chapter 12). However, helping such people to achieve their potential, and also treating the distress that has warranted a diagnosis in the first place, is clearly desirable. To do this effectively it is necessary to understand the causes of the problems they face.

Although far from complete, the process of unravelling the proximal\(^1\) (nearest: most immediate) neuropsychological causes of autism has already contributed to establishing rationales for psychosocial, behavioural and educational intervention (see Chapter 12). When the intermediate neurobiological causes – i.e. abnormalities of brain structure, chemistry and function – in autism are better understood, it is certain that pharmacological and possibly other physical treatments will be developed that effectively target specific aspects of behaviour, possibly in specific subgroups of individuals. Identification of etiological causes of autism is important for prevention. The identification of environmental factors that may be involved is particularly urgent and potentially important, because environmental factors may be modifiable, offering the prospect of reducing the incidence of ASD and associated problem behaviours. Understanding the genetic abnormalities that can cause, or make an individual vulnerable to, autism may also contribute to prevention in the future.

There are also theoretical spin-offs from investigations into the causes of ASD. In particular, such research should contribute to what is known about the links between brain and behaviour in typical development, and to understanding links between the functions of particular genes in shaping normal brain development. These advances in knowledge should have practical applications for the health and wellbeing of many groups of children.

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\(^1\)Words or phrases in bold type on first occurrence can be found in the Glossary.
COMPLICATIONS AND SIMPLIFICATIONS

Identifying the causes of autism is an exceedingly complicated and difficult undertaking, for the following reasons.

- Autism is a complex condition affecting very many aspects of behaviour. It is also very variable in the ways in which it is manifested in different individuals and at different stages of development, as emphasised in earlier chapters.
- There are three broad levels at which accounts of the causes of autism-related behaviour may be pitched, as identified in the second paragraph of this chapter. Not only must the causes of manifest behaviours associated with ASD be understood at each of these levels separately. In addition, ways in which the different levels of explanation relate to each other must also be identified, as shown by the arrows in Figure 6.1.
- There is no simple explanation of autism: the causes of autism-related behaviours are complex, cumulative and interactive.

In the next three subsections more will be said about each of these sources of difficulty in turn, with an explanation of the strategies to be used to simplify the material to be presented in subsequent chapters in Part II.

Identifying a Realistic Agenda

In the long term it may be possible to explain all the myriad bits of the puzzle which is autism: why age of onset varies; why young children with autism often have larger-than-usual heads; why they are often fussy eaters; why they so commonly

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**Figure 6.1** *Causal links that must be established*
What Causes Autism?

develop epilepsy; the relationship between autism and exceptional achievement; why autism has not been removed from the gene pool by natural selection; and so on and so forth – an impossible agenda given the limitations of current knowledge. In the shorter term, therefore, the priority is to understand the causes of the diagnostic behavioural characteristics:

• Socio-emotional-communicative impairments and anomalies (SEC impairments).
• Restricted and repetitive behaviours, including sensory-perceptual anomalies (RRBs).

It is also important to understand the causes of the two most common and jointly debilitating specifiers:

• Learning disability.
• Language impairment.

Subsequent chapters in Part II will focus on this simplified agenda.

Keeping the Explanatory Levels Apart and Putting Them Together

In subsequent chapters in Part II, theories and evidence relating to causes of ASD-related behaviours will be considered at each explanatory level separately. Specifically, Chapter 7 considers what is known about the etiology (distal or ‘root’ causes) of the four sets of ASD-related behaviours listed under the bullet points above. Chapter 8 considers the neurobiology (‘brain bases’) of these ASD-related behaviours. Chapters 9 and 10 summarise current theory and evidence relating to the neuropsychology (proximal or ‘immediate’) causes of SEC impairments and RRBs, and of learning disability and language impairment. Links between explanatory factors at different levels of explanation are referred to where these have been reliably demonstrated.

Most space is given to explanation at the neuropsychological level for various reasons. First, more is known about the neuropsychology of ASD-related behaviours than about either the etiology or neurobiology underlying these behaviours. In the second place, educational, psychosocial and behavioural treatment methods should ideally be based on a detailed understanding of the neuropsychological processes underlying the patterns of ability and disability that typically occur in people with ASD. In this connection, it might be argued that understanding the neurobiology and especially the etiology of ASD could lead to prevention or cure, and that these aims should take priority over improvements to educational or behavioural treatments. However, the desirability of eliminating autism from the population may be questioned (see Chapter 12), whereas interventions that increase day-to-day competencies and decrease maladaptive behaviours are unarguably desirable. A third, purely pragmatic reason for the imbalance between space given to neuropsychology as opposed to other levels of explanation is that etiology and neurobiology both
involve specialised knowledge too detailed to include in this book. Only the most basic information about these subjects is therefore covered in Chapters 7 and 8, with references cited where fuller accounts can be found.

Simplifying the Search for Causes

A diagram representing the causal links that may be implicated in any complete explanation of autism, let alone of autistic behaviours in any one individual, would show a dense tangle of cause–effect links. Major sources of this complexity are outlined below, before ways of simplifying the search for causes are suggested.

Sources of complexity

No single causal pathway  Early attempts to explain autism focused on explanations implicating a single causal factor, or ‘single common pathway’, at one or other level of explanation. Some of these early theories were mentioned in Chapter 1. These, and other examples of single common pathway theories, are shown in Box 6.1.

Box 6.1 Some single common pathway theories

<table>
<thead>
<tr>
<th>At the etiological level</th>
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<tr>
<td>Genetically determined oversensitivity to oxygen (as administered at birth) (Rimland, 1964).</td>
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<table>
<thead>
<tr>
<th>At the neurobiological level</th>
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<tr>
<td>Abnormal function of the reticular activating system in the brain (Hutt et al.1964).</td>
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<tr>
<td>Abnormal function of the vestibular system in the brain (Ornitz &amp; Ritvo, 1968).</td>
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<tr>
<td>Basal ganglia and mesial frontal abnormalities (Damasio &amp; Maurer, 1978).</td>
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<table>
<thead>
<tr>
<th>At the neuropsychological level</th>
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<tbody>
<tr>
<td>Lack of innate capacity for emotional relatedness (Kanner, 1943).</td>
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<tr>
<td>Faulty conditioning (Ferster, 1961).</td>
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<td>Cold or neglectful parenting, especially by the mother (Bettelheim, 1967).</td>
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<td>Severe language disorder (Churchill, 1972).</td>
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<td>Defective understanding and use of symbols (Ricks &amp; Wing, 1975).</td>
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<tr>
<td>Defective sequencing ability (Tanguay, 1984).</td>
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<tr>
<td>Impaired theory of mind (Baron-Cohen, 1989).</td>
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Single common pathway explanations of autism are attractive because they are parsimonious. However, single factor theories of ASD are undermined by evidence relating to what has been called the broader autism phenotype (BAP) or lesser
variant autism (Pickles et al., 2000; Wilcox et al., 2003; Dawson et al., 2002). This evidence shows that it is quite common for relatives of individuals with ASD to show signs of one or another kind of autism-related behaviour, in isolation from any other signs. A sibling, for example, may be something of a loner, but is not obsessive or routine-bound, and has good language. A father may be an avid collector of something, but at the same time sociable and articulate. An aunt may have had speech and language therapy as a child, and was slow to learn to read, but she has no problems with personal relationships and is not one-track minded or routine-bound. Evidence that gave rise to the concept of a broader autism phenotype, or lesser variant autism, shows that the SEC impairments and RRBs that define ASD – also vulnerability to language/learning impairments – are dissociable: any one of them can occur without the others. It follows that they must have at least partly different causes (Happé et al., 2006).

Many-to-one and one-to-many  Life would be easier for those seeking to understand the causes of autism if each facet of autism-related behaviour had a single, clear-cut explanation at each level – etiological linking to neurobiological linking to neuropsychological. Unfortunately this is not the case. For example, impaired mentalising may be a critical cause of SEC impairments, as is suggested in Chapter 9. However, obsessive interests, abnormal sensory perception, and poor language comprehension, if present, may also contribute. Similarly, RRBs (including sensory-perceptual anomalies) may possibly result mainly from poor control of arousal levels, as is also discussed in Chapter 9. However, anxiety, maladaptive learning, or comorbid obsessive-compulsive disorder may also contribute. The phrase ‘many-to-one’ (coined, I think, by Uta Frith) neatly captures the fact that each facet of autism-associated behaviour has many contributory causes (see Figure 6.2a).

At the same time, a single causal factor can have multiple effects. For example (and again considering only the neuropsychological level of explanation), impaired mentalising is not only a critical cause of SEC impairments but also an important cause of impaired sense of self and a cause of certain anomalies of language. Hence the phrase (also Frith’s) ‘one-to-many’ (see Figure 6.2b).

Multi-directional causal links  It might be assumed that links in the chain of causes and effects always travel ‘upwards’, as suggested by the vertical arrows in Figure 6.1: that is to say, from root causes to brain bases to the immediate neuropsychological causes of manifest behaviour. Unfortunately, yet again, the reality is not in fact so simple: as well as ‘forwards’ cause–effect links, there are also ‘sideways’ causal links. For example, at the neuropsychological level, impaired ability to recognise faces aggravates a pre-existing lack of interest in, and lack of attention to, faces, which then feeds backwards into the original recognition impairment, making it worse than it originally was. Similarly, at the neurobiological level, a congenital abnormality in a subcortical structure that normally sends information to certain cortical structures will affect the development and function of those cortical structures. At the etiological level it is likely that genes underlying vulnerability to autism are interactive as well as cumulative.
Individual differences Many of the causal factors mentioned above as possibly contributing to autistic behaviour do not apply to everyone with autism. The whole complex of interactive causal factors contributing to autism-related behaviours in any one individual will therefore be unique.

Simplifying the search for causal factors

In view of the complexity and diversity of causal pathways to autism in its various forms, let alone in unique individuals, the following simplifying strategy will be used in subsequent chapters in Part II.

Only the most important causal factors at each level of explanation will be considered. These will be referred to as ‘critical causes’ or ‘critical causal factors’. Such critical factors are those that may be necessary and/or sufficient to cause a certain set of autism-related behaviours to develop. By ‘necessary’ is meant that a particular causal factor must be present for a particular effect to occur. For example, the AIDS virus is a necessary cause of AIDS: people do not develop AIDS unless exposed to the virus. It is also a ‘sufficient’ cause, because nothing else is needed for AIDS to occur. By contrast, there is no necessary cause of an ailment such as toothache. Rather, toothache can occur for a number of reasons such as an abscess, caries or breaking a tooth, any of which can be sufficient for it to occur, but none of which is a necessary cause – because there are alternatives.
What Causes Autism?

It is safe to assume that there are certain necessary and/or sufficient causes of each of the defining impairments in ASD, and of the major specifiers. In what follows, these critical causes will be explored. The contributory role of factors which are neither necessary nor sufficient to explain facets of autism-related behaviour may be mentioned, but will not be discussed in detail. Parents, teachers, therapists and others in day-to-day contact with specific individuals on the spectrum do not, of course, have the luxury of simplifying in this way. For them, in order to understand and respond appropriately to a particular behavioural difficulty or stumbling block to learning, it may be necessary to unravel a whole complex of causes.

ASSESSING THE MERITS OF CAusal THEORIES

At present no one knows for certain what causes autism, whether in terms of root causes, brain bases or neuropsychological anomalies. It is essential, therefore, to keep an open mind, but at the same time to be critical in the best sense of that word, when considering theories of the possible causes of autism. In what follows, some points to bear in mind when critically appraising the merits of any particular theory are suggested.

Some Points to Bear in Mind

Recent Does Not Necessarily Mean Right

The list of ‘single common pathway’ explanations of ASD, outlined in Box 6.1, shows that explanatory theories come and then usually go, to be superseded by other theories, which are superseded in their turn. Each of the early theories was among the most recent when first presented, but most were subsequently rejected. There is no reason to suppose that this process has entirely ceased, although with the benefit of 50–60 years of research, recent theories are more securely evidence-based than earlier theories.

Old does not necessarily mean wrong

However, old theories should not be entirely discounted. Sometimes a theory is largely ignored when first proposed, but is recognised years later as having been ahead of its time. For example, in 1943 Kanner proposed that autism derives from ‘an innate inability to form the usual biologically provided affective contact with people’. This theory was ignored for several decades because, first, psychoanalytic explanations of autism dominated the literature; then behaviourist explanations; then explanations based on thinking of the brain as if it were a computer – ‘information processing’ models. Fifty years after it was first proposed, Kanner’s theory that ASD is an innate, brain-based condition was revived (by Hobson, 1993, in particular) and is now seen as consistent with mainstream
contemporary theories, as will become clear in Chapter 9. Similarly, the theory that dysregulation of arousal levels may explain RRBs in ASD, also discussed in Chapter 9, was first proposed by Hutt et al. in 1964. Consider, also, one of the earliest attempts to establish a set of diagnostic criteria for ASD, namely ‘Creak’s Nine Points’ (see Box 1.2). These included ‘abnormal response to perceptual stimuli’, now reinstated within diagnostic criteria after many years of absence; and ‘apparent unawareness of personal identity’, a characteristic confirmed by recent research, but little commented on in the intervening period.

It is also important to appreciate that theories receiving even temporary recognition are not plucked out of thin air: they are all based at the very least on observation and experience, and usually also on evidence from research. So, for example, Bettelheim’s psychoanalytic theory was based on evidence that mothers’ relationships with their young autistic child are abnormal. It was later realised that this is not because these mothers are by nature cold and uncaring, as originally suggested; but because it is difficult, often unrewarding, and sometimes distressing to try to establish a loving, interactive relationship with a young child with autism, especially if the nature of the child’s problem is not at first understood. It is now widely recognised that parents and other main carers of young children with ASD may need support and advice as to how best to establish a loving relationship with their difficult child, and perhaps how to cope with their own feelings of disappointment and frustration.

Who says?
The people who know about autism in vivid detail are people who are themselves autistic, their families, carers, and others in day-to-day contact with them. Any theory that is not informed by, and consistent with, the experience of the majority of those closely involved with autism is not likely to be correct. Research findings generally confirm what parents, teachers, and high-functioning people themselves know. Research may clarify or extend what ‘insiders’ know, while rarely conflicting with it (and when it does, it is generally the research that needs to be looked at again). Equally, anecdotes or observations from insiders’ accounts are often the seed corn for a particular theoretical hypothesis. For example, the first study of pretend play with which I was involved, carried out at a time when it was commonly said that children with autism cannot pretend, was motivated by a teacher telling me that a not very able girl with autism had returned from a visit to a stately home and trailed a coloured scarf behind her, saying ‘bird’: she had seen peacocks on her day out!

People with ASD and those who care for and work with them may not, however, be in a good position to see the bigger picture: they see one, or a few, of the trees in vivid detail, but not the whole wood made up of many and diverse trees. Researchers and theoreticians have their eyes on the wood as a whole. Reliable evidence about autism in general must therefore come from research studies, and these must be properly conducted and properly interpreted. Well-conducted, appropriately interpreted research studies are most likely to be found in journals that use the vetting process of peer review. Research reports that are published without peer review constitute what is known as the grey
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literature. It is wise to be aware of the theories and evidence being discussed in the grey literature: there is rarely smoke without fire, and much of the grey literature is written by parents or clinicians who have detailed experience of autism. However, the more reliable source of evidence and theories concerning the causes of autism is the peer-reviewed journals. All the research studies referred to in subsequent chapters are published in journals with a high standard of peer review.

Criteria for Judging the Strength of Theories

When judging the explanatory power of theories relating to autism, the following criteria should be borne in mind.

Specificity criterion  According to this criterion, if a theory proposes that critical factor x is necessary and sufficient to cause a particular facet of autism, then factor x (whether at the etiological, neurobiological, or neuropsychological level of explanation) must be specific to, that is to say, unique to, people with ASD. If factor x is not specific to individuals with ASD there is a problem of explaining why other individuals, to whom factor x also applies, do not show autistic behaviours. For example, it was proposed for a time that an impairment of explicit theory of mind (see Chapter 3) is the critical cause of the socio-emotional-communicative impairments diagnostic of autism. However, typically developing children below the age of 3 years 6 months, as well as many learning disabled individuals, also fail tests of explicit ToM, but are not autistic. This impairment cannot, therefore, be the necessary and sufficient cause of SEC impairments in ASD.

Universality criterion  This criterion states that if a theory proposes that critical factor x is a necessary (even if not sufficient) cause of one of the defining or additional shared features of autistic behaviour, then factor x must be shown to occur universally in all individuals with ASD. If not universally present, then factor x cannot logically be a necessary cause of that facet of autism-related behaviour. For example, if it is proposed that impaired mindreading is a necessary cause of SEC impairments in autism, then it must be shown that all individuals with ASD have impaired mindreading.

Primacy criterion  The primacy, or ‘causal precedence’, criterion requires that factor x must occur at an earlier developmental stage than the abnormality that factor x is supposed to explain. This is because it is a law of nature that causes precede effects. The original much-hyped 1980s/1990s ‘impaired theory of mind’ explanation of socio-emotional-communicative impairments in ASD failed on this criterion, because SEC impairments occur in infants with ASD usually within the first 30 months of life. Because the ability to pass explicit ToM tests does not mature in typically developing children until much later than this, lack of an explicit ToM cannot explain the early signs of SEC impairments in autism.
SUMMARY

It is important to understand the causes of autism in order to progress towards better treatments and possible prevention of debilitating forms of ASD at some future time. Understanding the causes of autism will also contribute to understanding brain development and brain–behaviour relations in typically developing children.

However, explaining autism is difficult, for numerous reasons. These include the fact that autism is a complex condition affecting very many aspects of behaviour. It is also very variable in the ways in which it is manifested in different individuals and at different stages of development. In addition, there are three broad levels at which accounts of the causes of autism may be pitched: the etiological or ‘first causes’ level; the neurobiological or ‘brain bases’ level; and the neuropsychological or ‘immediate causes’ level. Not only must the causes of autism be understood at each of these levels separately: the ways in which etiological factors give rise to neurobiological abnormalities and anomalies, which in turn cause neuropsychological abnormalities, must also be identified. Moreover, there is no single critical cause, no ‘single common pathway’, at any of the three levels of explanation. Instead, it is certain that the causes of autism-related behaviours are complex, cumulative, interactive, and to some extent individualistic.

Some ways of simplifying the explanatory task are outlined, to be used in subsequent chapters of Part II of this book. These include the following: focusing only on the universal or most common behavioural characteristics; focusing on the identification of causal factors at each level of explanation separately; and seeking to identify only the main, or most critical, cause(s) of each of the universal or very common behavioural impairments, ignoring more minor contributory causes.

Finally, it is important to maintain a critical attitude when considering the relative merits of particular theories of the causes of autism. The most recent theory is unlikely at this stage of knowledge to be completely correct; equally, older theories should not be automatically discounted. The research evidence cited in support of a particular theory should be carefully considered before being accepted, and the explanatory adequacy of each theory judged on the criteria of specificity, universality and primacy.