Developmental Disorders of Language Learning and Cognition

Charles Hulme and Margaret J. Snowling

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Understanding Developmental Cognitive Disorders

John, Peter, and Ann are three 7-year-old children. John's parents and teachers have concerns about his progress in learning to read. John is generally bright and understands concepts well. Formal testing showed that he had a high IQ (120) with somewhat higher scores on the performance than the verbal scales of the test. John could only read a few simple words on a single word-reading test – a level of performance equivalent to a typical 5½-year-old child. John does not know the names or sounds of several letters of the alphabet. Verbally John is a good communicator, though he does show occasional word-finding problems and occasionally mispronounces long words. John is a child with dyslexia.

Peter is also a bright little boy (IQ 110, but with markedly lower scores on the performance than the verbal subtests). He has made a very good start with learning to read, and on the same test given to Peter he read as many words correctly as an average 8-year-old child. Peter has severe problems with games and sport at school, particularly with ball games. He is notably ill-coordinated and frequently drops and spills things. He has very serious difficulties with drawing and copying, and his handwriting is poorly formed and difficult to read. Peter has developmental coordination disorder.

Ann is a socially withdrawn child. She avoids interacting with other children in school whenever she can. She is sometimes observed rocking repetitively and staring out of the classroom window. Ann's communication skills are very poor, and she appears to have quite marked difficulties understanding what is said to her, particularly if what is said is at all abstract. When an attempt was made to give Ann a formal IQ test, testing was discontinued because she refused to cooperate. The few items she did complete suggested she would obtain a very low IQ score. Ann is fascinated by cars and will spend many hours cutting out pictures of them to add to her collection. Ann is a child with autism.

These three cases of 7-year-old children illustrate some of the varied cognitive problems that can be observed in children. In this book we will attempt to provide a broad survey of the major forms of cognitive disorder found in children, and lay out a theoretical framework for how these disorders can best be understood.

Understanding these disorders, in turn, holds prospects for how best to treat them. Our approach to these disorders is from a developmental perspective, by which we mean that a satisfactory understanding of these disorders needs to be informed by knowledge of how these skills typically develop. Most of the explanations we consider in the book will focus on the cognitive level: a functional level dealing with how the brain typically learns and performs the skills in question. Wherever possible, however, we will relate these cognitive explanations to what is known about the biological (genetic and neural) mechanisms involved in development. The interplay between genetic, neural, and cognitive explanations for behavioral development is currently an area of intense activity and excitement.

Some Terminology for Classifying Cognitive Disorders

In this book we will consider a wide range of developmental disorders that affect language, learning, and cognition. The disorders considered include those affecting language, reading, arithmetic, motor skills, attention, and social interaction (autism spectrum disorders). There are a number of features that are shared by the disorders we will discuss: they all occur quite commonly and have serious consequences for education, and thereafter for well-being in adulthood. There is also good evidence that all these disorders reflect the effects of genetic and environmental influences on the developing brain and mind.

To begin with it is important to distinguish between specific (or restricted) difficulties and general difficulties. Specific difficulties involve disorders where there is a deficit in just one or a small number of skills, with typical functioning in other areas. General difficulties involve impairments in most, if not all, cognitive functions. Terminology in this field differs between the UK and the USA; we will consider both here, but we will use primarily British terminology in later sections of the book.

In the UK a selective difficulty in acquiring a skill is referred to as a "specific learning difficulty." The term learning difficulty makes it clear that skills must be learned; specific means that the difficulty occurs in a restricted domain. Dyslexia is one of the best known and best understood examples of a specific learning difficulty. Children with dyslexia have specific difficulties in learning to read and to spell, but they have no particular difficulty in understanding concepts and may have talents in many other areas such as science, sport, and art. In the USA (following DSM-IV, the *Diagnostic and Statistical Manual of Mental Disorders* of the American Psychiatric Association) such specific difficulties are called learning disorders.

Specific learning difficulties can be contrasted with general learning difficulties (or, in US terminology, mental retardation). General learning difficulties involve difficulties in acquiring a wide range of skills. People with the chromosomal abnormality of Down syndrome, for example, usually have general learning difficulties and typically have problems in mastering all academic skills and with understanding in most domains. In this book we will focus upon specific learning difficulties.

In practice, the distinction between specific and general learning difficulties is often based on the results of a standardized IQ test. IQ tests (or measures of general intelligence) are highly predictive of variations in attainment in all manner of settings. The average IQ for the population is 100 (with a standard deviation of 15 points). In the UK people with IQ scores between 50 and 70 are referred to as having moderate learning difficulties, and people with IQ scores below 50 are said to have severe learning difficulties. US terminology distinguishes between mild (50–70), moderate (40–50), severe (25–40), and profound (IQ below 20) mental retardation. Often the diagnosis of a specific learning difficulty is made only in cases where the child achieves an IQ score in the average range (perhaps an IQ of 85 or above).

Operationally the distinction between specific and general learning difficulties is therefore quite clear: children with specific learning difficulties typically have average or near to average IQ scores, while children with general learning difficulties have IQ scores below 70. Conceptually, however, the distinction is probably a bit more slippery. It is important to appreciate that there is a continuum running from the highly restricted deficits found in some children (e.g., a child with a severe but isolated problem with arithmetic), to more general difficulties (e.g., a child with severe language difficulties who has difficulties both with understanding speech and expressing himself in speech), to very general difficulties (a child with an IQ of 40, who is likely to have problems in reading and spelling, as well as spoken language, together with a range of other problems including problems of perception, motor control, and general conceptual understanding). One aim of this book is to convey an appreciation of how studies of children with different types of learning difficulties have contributed to an understanding of how a range of different brain systems are involved in learning. The range of learning difficulties that occurs ultimately helps us to understand how the developing mind is organized and how the skills that are impaired in some children are typically acquired.

Levels of Explanation in Studies of Developmental Cognitive Disorders

What form of explanation can we hope to achieve for developmental cognitive disorders? It is important to distinguish between the different levels of explanation that are possible. Morton and Frith (1995) have laid out very clearly the logic and importance of distinguishing the different levels of explanation that are needed for understanding developmental disorders. They show how it is essential to consider three major levels of explanation: biological, cognitive, and behavioral. At each of these levels underlying processes (in the child) interact with a range of environmental influences to determine the observed outcome.

We can illustrate the role of different levels of explanation with reference to conduct disorder, a disorder of socio-emotional development that we will not deal with further in this book. Conduct disorder is a disorder where there have been advances in understanding at several different levels recently (Viding & Frith, 2006) and it is

therefore a good example to illustrate the different levels of explanation involved in the study of developmental disorders. Conduct disorder is defined in DSM-IV as persistent antisocial behavior that deviates from age-appropriate social norms and violates the basic rights of others (American Psychiatric Association, 1994); alternative terms sometimes used for this disorder include antisocial behavior and conduct problems. A model for one aspect of conduct disorder – reactive aggression – proposed by Viding and Frith (2006) is shown in Figure 1.1 below.

This model represents processes operating at the biological, cognitive, and behavioral levels of explanation. It appears that at the biological level specific differences in genes that regulate the action of the neurotransmitter serotonin are important in giving rise to a predisposition to commit acts of violence. More specifically, different variants (alleles) of a gene coding for monoamine oxidase inhibitor A (MAOA) have been identified, with either high (MAOA-H) or low activity (MAOA-L). Research has suggested that having the MAOA-L gene may predispose an individual to display violent behavior but only if they experience maltreatment in childhood (Caspi et al., 2002). (This is a very important finding since it provides an example of gene–environment interaction; neither having the gene nor being maltreated alone may be sufficient but both factors together give a greatly increased risk of developing

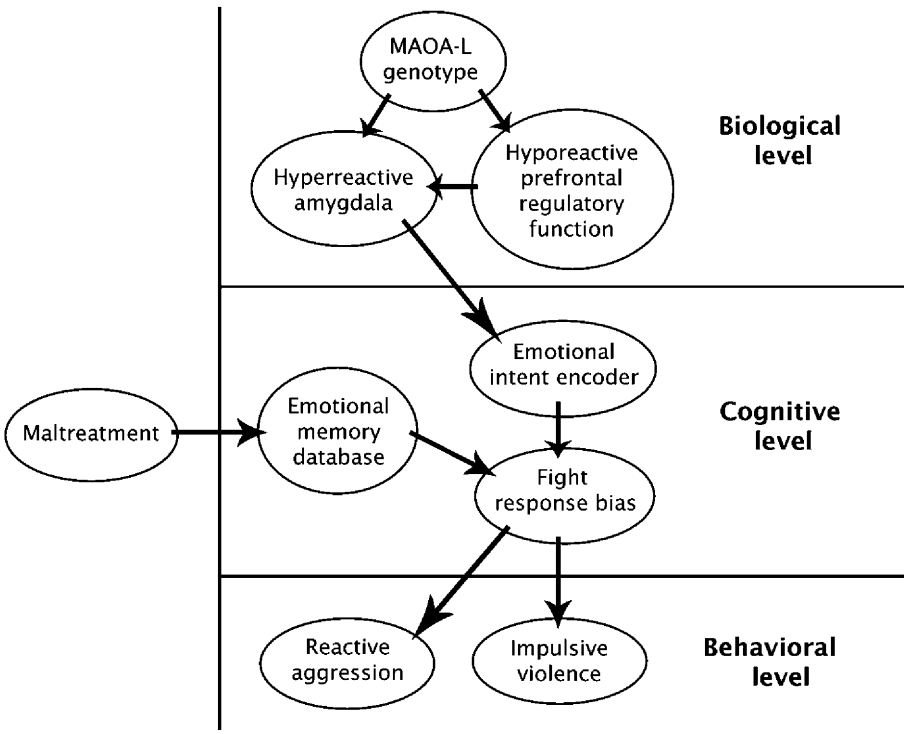

Figure 1.1 A causal model of the potential gene–brain–cognition–behavior pathways from MAOA-L to reactive aggression. (Adapted from Viding, E. & Frith, U. Genes for violence lurk in the brain. Commentary. *Proceedings for the National Academy of Sciences, 103*, 6085–6086. Copyright (2006) National Academy of Sciences, USA.)

conduct disorder.) These genetic and environmental risk factors in turn appear to operate on the development of brain systems concerned with the regulation of emotion. In particular it is thought that the MAOA-L gene may be associated with the development of hyperresponsivity of the amygdala during emotional arousal coupled with diminished responsivity of areas of the prefrontal cortex that normally play a role in regulating such emotional responses. This pattern of brain dysfunction might be seen as providing the biological basis for reacting excessively emotionally and violently when provoked by certain environmental conditions (in everyday terminology, losing control or "losing it" when provoked).

Viding and Frith suggest that these brain differences express themselves at the cognitive level via a mechanism called an emotional intent encoder, which in turn is associated with a bias to fight. Interestingly, in this model, Viding and Frith explicitly propose that the interactive effects of childhood maltreatment operate at a cognitive level by leading to the creation of many emotionally charged memory representations. This is an interesting and testable hypothesis, but of course such effects may also operate at a biological level as well as, or instead of, at the cognitive level.

The final level in the model is the behavioral level, where the fight response bias mechanism may lead to reactive aggression (fighting when provoked) as well as impulsive violence.

A complete explanation of any disorder will involve at least three levels of description. For one aspect of conduct disorder – reactive aggression – genes appear to contribute powerfully to the risk of developing the disorder in interaction with specific environmental experiences (maltreatment) in childhood. It appears that these genetic effects in turn affect the development of brain circuits concerned with the experience and regulation of emotion, perhaps particularly anger, which, in interaction with memories of previous experiences associated with violence, may lead to a bias toward fighting (rather than running away or being afraid). At a behavioral level, this bias toward a fight response may lead to the observed profile of responding violently when provoked and occasionally committing unprovoked, impulsive acts of violence.

Morton and Frith (1995; Morton 2004) argue that it is useful to make explicit diagrams of these sorts of theoretical explanations, using an approach they term causal modeling. The Viding and Frith diagram (Figure 1.1) is an example. It is important to note that the arrows in such a diagram represent hypothetical causal links. According to this model, a genetic difference causes a brain difference (abnormality), which in turn causes cognitive (emotional) deficits, which in turn cause the observed behavioral patterns (a propensity to violence). Note that within this framework environmental effects can be thought of as operating at each level. So, for example, a virus or early brain injury might also lead to the brain abnormality underlying the emotion control problem, and the effects of positive experiences (a nurturant nonaggressive parental style) might prevent the development of the emotion regulation deficits. Some forms of treatment (teaching anger management strategies) might also have effects on the behavioral level (inhibiting violent outbursts) without having a direct effect on the cognitive level (the person may still feel angry and feel the urge to lash out, but develop ways of controlling such feelings).

It is important to emphasize that all three levels of description are useful, and each helps us to understand the disorder. While links can and should be made between these different levels of explanation, we cannot reduce or replace one level of explanation with a lower level. The cognitive level of explanation (emotion encoding) cannot be replaced by a neural explanation (problems with the amygdala). We would note here that we have followed Morton and Frith's terminology by referring to the level between the brain and behavior as "cognitive." This might seem too narrow a term because cognition essentially refers to thought processes. We will stick with this term for the moment, though in some of the disorders we consider later (as well as in the case of conduct disorder) this terminology might usefully be broadened to consider other forms of mental processes, particularly emotional and motivational processes, that probably cannot simply be reduced to cognition. The point, however, is that we need a level of "mind" or "mental process" as an intervening level of explanation between brain and behavior. We would also argue, in light of recent advances in our understanding of developmental disorders, that the causal model presented in Figure 1.1 is too unidirectional to capture the truly interactive nature of development. It is also necessary to postulate causal arrows running "backwards" from lower levels to upper levels. This at first seems counterintuitive, but some examples help to explain why it is necessary.

Can changes at the behavioral level alter things at the cognitive level? Almost certainly yes. If we take the example of teaching anger management strategies mentioned above, it may be that such training will work by modifying the cognitive mechanisms associated with emotional encoding; seeing a person grin could be interpreted simply as showing that they were happy rather than indicating they are intending to insult you. Do such changes at the cognitive level depend upon changes in underlying brain mechanisms? Again it would seem likely that they do. Connections between nerve cells may be modified by experience and this in turn will result in lasting structural and functional changes in the circuits responsible for encoding and regulating emotion.

Finally, and perhaps most surprisingly, we can consider whether changes at the behavioral and cognitive levels can affect things at the genetic level. Most people would probably doubt this proposition. Our genetic makeup is fixed (we inherit our DNA at conception and experiences are not going to alter it), this is true, but there is evidence that experiences can alter the way genes are expressed. Genes (genes are sequences of base pairs in DNA) do not regulate development directly. Rather, genes control the production of messenger RNA (mRNA), and mRNA in turn controls the production of proteins in cells. Furthermore, mRNA molecules degrade quickly so that if more of a protein is needed the cells concerned have to keep manufacturing more mRNA. Changes in the rate at which a gene produces mRNA will therefore result in changes in the rate at which the protein coded is produced in a cell. The levels of regulation in cells, as currently conceptualized by molecular biologists, are shown in Figure 1.2. Once again, in this diagram there are different levels of explanation: the genome (the genes that consist of sequences of base pairs in DNA), the transcriptome (the mRNA produced under the control of the base sequences in the DNA), the proteome (the proteins produced under the control of mRNA), the

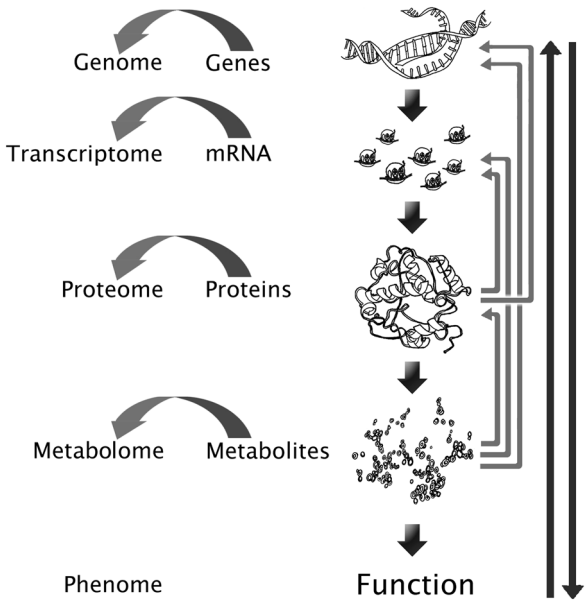

Figure 1.2 Diagram showing the complexities of genetic mechanisms. There are potentially numerous interactions at each level, as well as bidirectional influences between levels. All these parameters may differ between different developmental stages or in different tissues of the body. (With kind permission from Springer Science and Business Media. *Metabolomics*, Metabolomics – the way forward, 1, 2005, p. 2, Goodacre, R., fig.a.)

metabolome (the products of proteins and other chemicals created by metabolism in the cell), and the phenome (the functioning of the cell within its environment in the body).

As shown in Figure 1.2, there are bidirectional arrows connecting these different levels (not a one-way arrow flowing from DNA to Function). One of the ways in which experiences may affect the expression of the genome is through the operation of control genes. Such control genes exist to control the operation of other genes by switching these other genes on or off (i.e., making genes either produce or stop producing mRNA). It now appears that such control genes may cause other genes to be switched off in response to changes in the internal and external environment. One remarkable example of such effects is shown by the observation that tweaking a rat's whiskers may cause changes in gene expression in the animal's sensory cortex (Mack & Mack, 1992). Similarly, when a songbird hears their species' song this experience may operate to change the expression of genes in the brain (Mello, Vicario, & Clayton, 1992). Thus, we need to accept that environmental effects may result in changes in the way genes are expressed. Such changes in gene expression may in turn result in long-lasting changes in the neural structures whose development is partly under genetic control (see Plomin, DeFries, McClearn, & Rutter, 1997, for more details).

In line with these findings from animals it has been shown that in human monozygotic (identical) twin pairs there are measurable differences in patterns of gene expression (differences in the genes that are active or being expressed). Furthermore, these differences in gene expression increase with age and tend to be greater for twin pairs who have lived apart for longer and who have experienced greater differences in lifestyle and health (Fraga et al., 2005). These effects clearly suggest that differences in experience produce different patterns of gene expression in people and that such differences may be responsible for differences in health and brain development that may have effects on behavior.

Summary

We hope that our discussion makes clear that the environment affects how our genetic makeup is expressed. The patterns of gene expression in cells will differ in different tissues and at different stages of development. The tissues most relevant for explaining differences in behavior are those in the nervous and endocrine (hormonal) systems. The most important point for the present argument is to appreciate that experiences may affect the processes involved in gene expression. Viewed in this way, the genome is not fixed in the way it operates throughout development. Rather, the genome receives signals from the environment that can turn genes on or off in different tissues of the body (including the brain). This means that differences in our experiences may well affect how genes that play a role in controlling brain development are expressed.

For most of this book we will be concentrating on explanations for developmental disorders that seek to relate observed impairments at the behavioral level to deficits at the cognitive level. We believe that such cognitive explanations are important and valid in their own right. A cognitive explanation of a disorder is essentially a functional explanation, couched in terms of how a particular skill is learned and performed, and in what ways this typical functioning is disturbed. Such an explanation is satisfying in its own right, and also has practical importance, in that it relates closely (though always indirectly) to how we can best assess and treat a disorder. This is not to say that biological levels of explanation are not also important. We will, where appropriate, cite evidence about the biological mechanisms underlying the cognitive level of explanation, particularly where such biological evidence places constraints on the types of cognitive explanation that are most viable. As has already been made clear from the brief account of research on conduct disorder above, there are two levels of biological mechanism that may be particularly relevant to the study of developmental cognitive disorders: genetic and brain mechanisms. We will consider very briefly the way in which these mechanisms are studied.

Genetic Mechanisms

There are two levels at which the genetic basis of a disorder can be studied. Population genetic studies examine the patterns of inheritance of a disorder across individuals.

Molecular genetic studies go beyond this and identify certain genes (DNA sequences) or gene markers that are associated with the development of a disorder. Both of these levels of analysis have been applied in the case of conduct disorder.

Population genetic studies relate variations in genetic association to degrees of similarity in the phenotype (observed characteristics). Basically, if a characteristic is inherited, people who are genetically similar to each other should also be similar to each other in that characteristic. One of the ways to get such evidence is from studies of twins. These studies make use of the fact that there are two different types of twin. Identical or monozygotic (MZ) twins develop from a single fertilized egg. Nonidentical (sometimes referred to as fraternal) or dizygotic (DZ) twins occur when two different fertilized eggs implant in the womb at the same time. MZ twins effectively share all their genetic material, whereas DZ twins will only share on average the same degree of genetic similarity to each other as any other pair of siblings. (DZ twins should, on average, share 50% of their segregating or polymorphic genes. These segregating genes are the coding sequences of DNA that differ between people and contribute to individual differences. Such segregating genes only account for a tiny proportion of our DNA: indeed it has been suggested that human beings share 98% of their genetic code with chimpanzees.) Twin studies often involve making comparisons between how frequently a disorder occurs in pairs of MZ and DZ twins. If both twins in a pair share the same condition, they are said to be concordant. Concordance rates should be higher in MZ, than DZ, twin pairs if genetic factors are important.

Concordance rates are only really useful when studying characteristics that are either present or absent. For example, if breast cancer were influenced by genetic factors, we would expect that the risk of pairs of MZ twins both contracting the disease would be higher than for pairs of DZ twins. However, as we shall see later in the book, for many cognitive disorders it is difficult to set precise cut-offs for whether a person has, or has not, got a disorder. This is because the disorders are best described as dimensional (so that individuals can have a disorder to varying degrees). Because of this we need a method of studying the degree of similarity between pairs of twins when the measures are quantitative dimensions rather than categories. Such a method was developed by DeFries and Fulker (1985). This method basically uses a form of regression equation to assess the influence of genetic factors on a characteristic. If genes are important in determining a continuous characteristic (such as height), MZ twins should be more similar to each other on that characteristic than DZ twins.

The degree of genetic influence on the development of a characteristic is expressed in terms of a heritability estimate. Heritability is concerned with quantifying the extent to which differences among people in a population reflect genetic differences. A heritability estimate of 0 would mean that genetic differences played no role in explaining the differences among people in a characteristic, while a heritability estimate of 1.0 would mean that genetic differences accounted entirely for the differences observed. In practice heritability estimates are usually intermediate in size but it is common for developmental disorders to show substantial heritability, meaning that genetic influences are important for their development. To return to the case of

conduct disorder, there is good evidence that genetic factors are important for its development. For example, Blonigen et al. (2005) reported a heritability estimate of approximately 50 for a measure of impulsive antisociality in a large twin sample, meaning that some 50% of the differences between people on this measure reflected genetic differences between people in the sample studied.

Molecular genetic studies try to identify the specific genes that may be responsible for the development of a disorder. Modern techniques allow the sequence of base pairs in an individual's DNA to be "read off" quite rapidly. The problem then becomes one of sifting the huge amount of data generated. It would not be appropriate to go into the details of these methods here. However, the basic approach is to try to identify DNA sequences that are shared by close relatives who both display a disorder but are not shared by other close relatives who do not have the disorder. Such studies involve sifting huge amounts of data and, rather than identifying specific genes, quite large DNA sequences (consisting of potentially many genes) may be identified. A group of genes that can be shown to correlate with the development of a complex quantitative trait (such as reading ability) is referred to as a quantitative trait locus (QTL). However, in some cases specific candidate genes have been identified that appear to be causally related to the development of a disorder. In the example of conduct disorder described above, one of the variants (alleles) of a gene coding for low activity of the monoamine oxidase inhibitor A (MAOA-L) appears to predispose an individual to display violent behavior (but only if they experience maltreatment in childhood).

The Causes of Development – Nature Working with Nurture

One of the oldest and most central debates in developmental psychology is about the role of genes (nature) and environment (nurture) as determinants of development. As we will see later in the book, there is overwhelming evidence that genetic factors are powerful influences on the origins of many developmental disorders. We take this conclusion to be established beyond any reasonable doubt. This is not the same as saying the disorders are innate, however.

Innate is defined in the *Shorter Oxford English Dictionary* as "Existing in a person (or organism) from birth … inborn … of qualities … (especially mental) opposite of acquired …" It is important to appreciate that the idea embodied in this definition is totally at variance with current thinking in genetics and developmental biology. The critical point is that genes contain information that serves to direct development, but all development takes place in an environment and information from the environment interacts with the genetic "blueprint" in complex ways. Development results from the interaction of genetic and environmental inputs – an idea referred to as epigenesis. Furthermore, according to the idea of "probabilistic epigenesis" (Gottlieb, 1992; Johnson, 1997), there may be bidirectional influences between different levels so that, for example, genes that help to specify aspects of physical development (including brain development) can in turn be reciprocally influenced by the structures they have helped to produce (see Figure 1.3). Similarly, and perhaps more obviously,

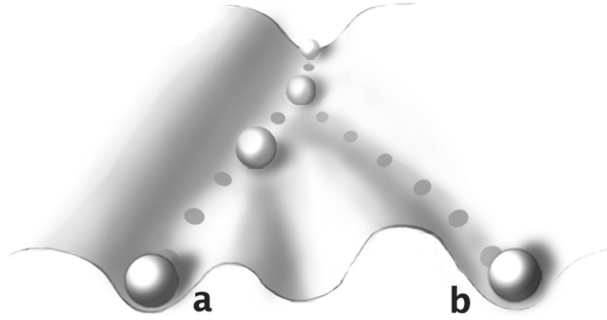

Figure 1.3 Waddington's epigenetic landscape is a metaphor for how gene regulation processes modulate development. Each of the marbles rolling down the hill represents a cell and different grooves in the landscape represent different trajectories that will result in different developmental courses and so different "end states" for a cell. Differences in the environment will play a role in determining the trajectory taken by a given cell. At a higher level we could think of the marbles representing whole organisms and again the end points of development will depend on both genetic and environmental influences.

learning (an influence from the environment) operates to modify structures in the brain that developed under genetic control and in turn may influence subsequent learning.

Development has to be seen as an extremely complex process that is characterized by change and interaction. All of the cognitive disorders we will consider in this book depend upon functional brain systems (brain systems that are defined by what they do) and it is simply not sensible to view these systems as arising directly and invariantly from information coded in the genes. In practice, performing any cognitive activity will depend upon one or more brain circuits, which comprise complex assemblies of many thousands of nerve cells communicating information between each other. Such brain circuits will develop under some degree of genetic influence but also as a product of learning from interactions with the environment.

Genes code for the production of proteins, which in turn have complex and at least partially indirect effects on the way physical structures such as the brain develop. Furthermore, as we have already noted, experiences may serve to switch on, or switch off, genes that are involved in controlling structural and functional aspects of brain development. In short, functional brain systems (brain circuits) develop as a result of complex interactions between genetic information and a range of environmental influences (where the environment includes many physical influences on development, such as temperature, nutrition, toxins, and radiation, as well as psychological experiences).

An acceptance that some aspect of development is under genetic influences does nothing to negate the importance of the environment. In relation to developmental disorders this can be illustrated by a well-known example. Phenylketonuria (PKU) is a genetic disorder that is controlled by a single gene. Children who inherit two such recessive alleles of this gene are unable to metabolize phenylalanine (an amino acid present in many foods) and this results in a build-up of this substance in the body

that damages the developing brain and causes general learning difficulties (mental retardation). However, PKU can be detected by a simple blood test (blood is taken in the heel prick test given to newborn babies) and provision of a special diet that is low in phenylalanine can prevent brain damage and the resulting learning difficulties from developing. A very clear discussion of the complex interplay between genetic and environmental influences on behavior is given by Rutter (2005b).

Brain Mechanisms

Genetic differences between people, in concert with environmental influences, determine the course of development, including development of the brain (epigenesis). In relation to developmental cognitive disorders it is likely that the problems we observe in different children will reflect both structural and functional differences in brain organization. In the last 20 years or so there has been an explosion of research concerned with understanding the relationships between brain, behavior, and cognition. Most of this research has focused on brain function, though it is also the case that some important work continues to examine the possible relationships between structural brain abnormalities and various forms of learning difficulties (Leonard, Eckert, Given, Berninger, & Eden, 2006)

Our ability to study the functional organization and operation of the brain while we are thinking has been transformed by the advent of brain imaging techniques. Positron emission tomography (PET) and functional magnetic resonance imaging (fMRI) are two techniques that have been used to study the patterns of neural activation occurring during ongoing cognitive tasks. Both PET and fMRI detect changes in blood flow in specific regions of the brain that arise during the performance of a task. When a brain region does work, it requires metabolic energy, which in turn requires extra oxygen and thus extra blood flow. Both of these techniques provide evidence for fairly slow-acting changes in brain activity and usually depend on averaging measurements from a number of trials in an experiment. However, the techniques give quite precise information about localization in the brain. The other methodological wrinkle is that we need to have a "baseline" against which to measure any putative increase in activation in a specific task. This therefore involves subtracting the levels of activation seen in a specific task from levels of activation seen in a similar task, preferably in a task that involves everything apart from the one component of an experimental task that we are particularly interested in. So, for example, activation might be compared in a condition where a subject sees and silently reads a sequence of words, and in another condition where exactly the same words are presented as pictures to be silently named. Areas of the brain that show increases in activation in the reading condition, compared to the picture condition, presumably are somehow specifically involved in processing written words (orthographic processing) and translating from orthography (print) to phonology (speech sounds). Details of the subtraction methodology become complicated, but the point is that imaging studies always involve some sort of inference to be made based on a comparison between closely matched tasks.

Electroencephalography (EEG) and magnetoencephalography (MEG) are two techniques that give better temporal (time-based) information about patterns of brain activity but poorer information about the localization of activity. EEG involves attaching electrodes to the scalp and measuring differences in voltage between the electrodes and how these voltage differences change across time. The timing of these voltage changes, which reflect patterns of firing from large sets of neurons in the brain, can be measured with millisecond (0.001 s) accuracy. One particularly useful EEG technique is event-related potentials (ERPs). To measure ERPs, EEG recordings are taken in response to a particular stimulus (or set of stimuli) and the results are averaged over many trials to identify consistent patterns of activity. MEG is a methodologically superior technique to EEG that also measures changes in neural activity in the brain. MEG measures the magnetic fields produced by the electrical activity in the brain by using superconducting quantum interference devices (SQUIDs), which are housed in a helmet-like enclosure that fits around the head (see Plate 1). Like EEG, MEG yields quite precise information about the timing of neural responses to stimuli, but it gives relatively crude information about the localization of activity in the brain. It seems likely that MEG will become a very valuable technique for studying brain activity, and combining MEG with fMRI recordings in the same individual provides the possibility of getting both localization and temporal information about patterns of brain activity.

Separable Systems in the Mind – Modularity and Development

Subsequent chapters in this book will consider what we know about the nature, origins, and treatments for a variety of developmental cognitive disorders. The fact that there is a wide range of somewhat specific developmental disorders (some children have difficulties with language, while other children have difficulties with the control of movement, for example) supports the idea that the mind has different systems (or modules) that are responsible for different functions (language and motor control in the case just cited).

The idea that the mind is a modular system (a system composed of separable subsystems) has a very long history that can be traced back at least as far as the ancient Greek philosophers (Arbib, Caplan, & Marshall, 1982). A slightly more recent, but now discredited, modular approach was represented in Gall's pseudo-scientific phrenology (see Figure 1.4). According to Gall the relative size of different brain regions (measured by feeling the shape of the skull!) could be used to infer characteristics of people, such as their "acquisitiveness" or "secretiveness." The idea of modularity has been brought to prominence in modern psychology by the work of Fodor (1983) and Marr (1983).

Studies of cognition in adults, and particularly studies in adult cognitive neuropsychology, have been dominated by an approach that sees the mind as a modular system. Cognitive neuropsychology seeks to develop theories about how the mind typically operates, by studying the disorders in mental (cognitive) processes that

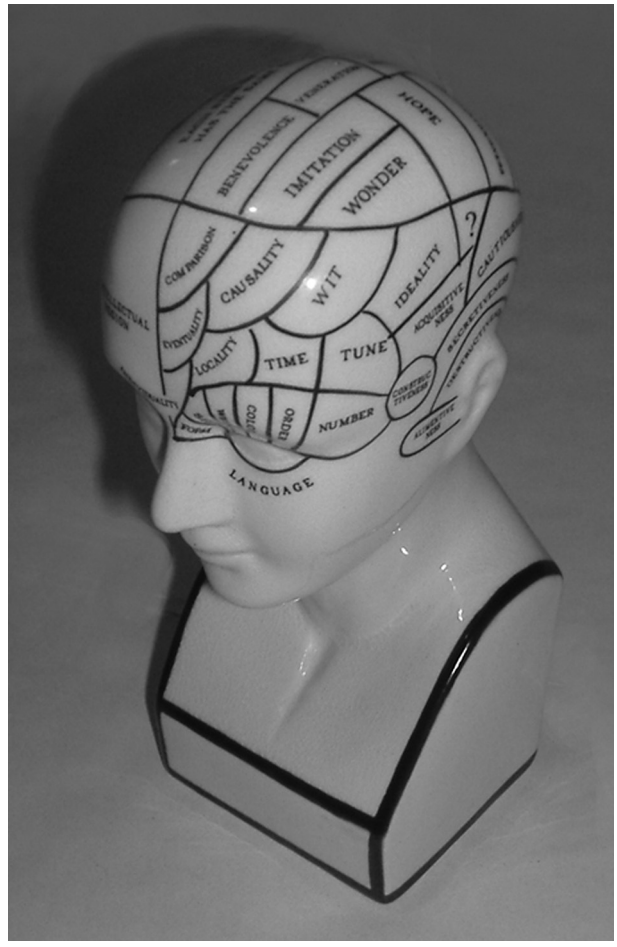

Figure 1.4 A phrenological head showing areas labeled with their supposed functions.

arise as a result of brain damage (see Shallice, 1988). A modular view sees the mind as composed of separate systems or modules, just as we might think of our bodies as being composed of different systems such as the circulatory, respiratory, and digestive systems. An analogy to convey the concept of modularity can be given by considering a computer (see Figure 1.5). A desktop computer usually consists of a number of interconnected components, some of which are physically separate (the monitor, keyboard) while others may be housed in the same box (the processor, hard disk, CD drive, sound card, video card, etc.). Problems in such a system can be easily identified, and rectified, by isolating or swapping components. To take a trivial example, if the monitor does not work, this may be due to a number of components (the monitor itself, the cable connecting it to the computer, or perhaps the video card inside the computer that generates the signals to control the monitor). By testing each of these components sequentially we can gradually identify the component that is causing the fault in such a system (though often such a process can be time consuming and frustrating!).

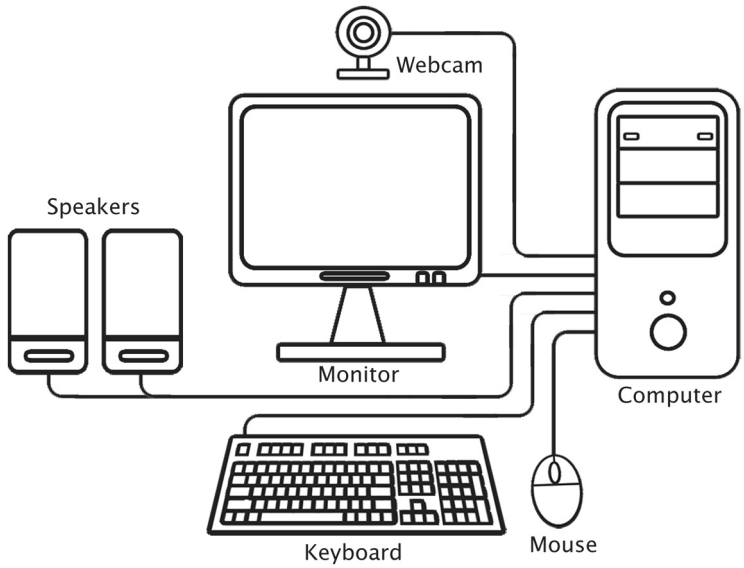

Figure 1.5 A computer as an example of a modular system.

In some ways, the studies we describe later can be thought of as analogous to this process of finding a fault in a computer system. If, for example, children with dyslexia perform tasks requiring them to isolate individual sounds in words very poorly, but perform as well as other children on analogous tasks requiring them to isolate shapes in complex visual displays, we might infer that the brain systems dealing with speech sounds are impaired in children with dyslexia, while other brain systems dealing with the perception of complex visual patterns are intact. We will, however, spend a great deal of time showing how understanding disorders of the developing mind is a much more complicated process than locating a fault in a computer system.

Cognitive neuropsychology in adults has made enormous progress by adopting an approach that seeks to understand the effects of brain damage as arising from impairments to separable cognitive systems that can be damaged independently as a result of brain injury. At the simplest level, modularity simply amounts to the claim that the mind consists of separate subsystems. To take an obvious example, there are separate systems responsible for vision and hearing in the brain. Damage to the primary visual cortex (at the back of the head, in the occipital lobe) results in areas of blindness, while damage to the primary auditory cortex (at the side of the head, in the temporal lobe) results in difficulties in discriminating the frequency of sounds (Tramo, Shah, & Braida, 2002). In these cases no one would wish to argue with the proposition that separate brain systems are responsible for the senses of hearing and vision, and that it is possible to get impairments in vision, without impairments in hearing, and vice versa. This, in the parlance of cognitive neuropsychology, would be an example of a double dissociation: patients with damage to the primary visual cortex have problems with vision, but hear normally; patients with damage to the primary auditory cortex have problems with hearing, but see normally. Double

dissociations have often been interpreted as providing critical support for modularity: the existence of separable, neurally independent, systems.

This example has been chosen deliberately to be clear and noncontroversial. We will make the reasonable assumption for the time being that the two patients described showed massive deficits on the visual and auditory tasks, but that each was completely normal on the nonimpaired task (one patient had a severe visual impairment, but completely normal hearing; the other had a severe auditory impairment, but completely normal vision). In such cases evidence of this sort can be related to a variety of other evidence (e.g., that the primary visual cortex receives input from cells in the retina of the eye, and stimulation of the eye by a flash of light results in neural activity in the primary visual cortex) to support a theory that the visual system is functionally and neurally separable from the auditory system. However, such very clear cases are the exception, even in studies of adults following focal brain lesions, and such distinctions become much harder to make once we move on to consider "higher" cognitive processes such as memory. Furthermore, as we shall see later, in studies of children with cognitive disorders such clear patterns of selective impairment are quite unusual (and this is an interesting point in its own right, to which we will return).

In reality the logic and practice of seeking to establish the existence of separate cognitive systems by looking for double dissociations is both controversial and complex and has been debated extensively (e.g., Coltheart & Davies, 2003; Dunn & Kirsner, 1988; 2003; Gurd & Marshall, 2003; Jones, 1983; Van Orden, Pennington, & Stone, 2001). There are both logical and statistical issues at stake in the debate about this issue. Logically, it seems reasonable to conclude that any given pattern of double dissociation might in principle be open to a variety of theoretical interpretations. Claims about separable processes will always depend upon having a clear theory about the processes concerned and finding converging evidence to support the idea of their separability (as in the case of converging evidence for the role of the visual cortex in vision described above).

At another level there are also purely statistical or methodological issues about how we need to measure behavior in order to establish dissociations between tasks (which is a prerequisite for trying to infer that the tasks depend upon dissociable mechanisms). In a typical case, the process of establishing an impairment in one domain, but not another, amounts to identifying what Chapman and Chapman (1973) referred to as a differential deficit. As these authors pointed out, identifying differential deficits depends critically upon the statistical properties of the measures used. In particular, the greater the true score (or reliable) variance in a test, the easier it will be to show that a clinical group is impaired on that test. True score variance increases as the reliability and the variance (the range of scores) of a test increase. The reliability of a test refers to the extent to which measurement is subject to error. The variance in scores from a test will vary with the relative difficulty of the test for the sample of people it is used with: the variance in test scores will decrease when tests are either too hard (tendency toward a floor effect) or too easy (tendency toward a ceiling effect). The statistical methods needed to identify differential deficits are well understood, though in practice these methods can be onerous and are rarely followed rigorously.