Part 1
Disorders of intellectual development: concept and epidemiology

CHAPTER 1

Disorders of intellectual development: historical, conceptual, epidemiological and nosological overview

Anthony Holland

There are different ways of thinking about the needs of people with disorders of intellectual development (DID); each has its place and none is perfect. The aim of this chapter therefore is to provide an overview of the various perspectives that those working in this field may use to orient themselves to the issues. In clinical practice, when seeing someone who has been referred, the starting point is to ask the question: What am I being asked to do? For the paediatrician and/or geneticist it may well focus on identifying whether a single major cause for a child's developmental delay can be identified. For a psychiatrist, clinical psychologist or community nurse it may be about identifying the reasons for, and treatment of, a particular constellation of problem behaviours. The task, through history taking, observation, examination and investigation, is to arrive at an understanding – a formulation – that then informs intervention through the integration of information about the individual within an accepted theoretical and conceptual framework that has been developed through research.

While DSM-5 uses the term 'intellectual developmental disorders' (American Psychiatric Association, 2013), the term generally used in this book is the one due to be used in ICD-11: 'disorders of intellectual development'. It is the latest in a long line of labels that have included a range of unacceptable and derogatory terms, from idiot, imbecile, feeble-minded and moral imbecile to mentally retarded, mentally handicapped and mentally subnormal, and, more recently, learning disabled and intellectually disabled. Many of these terms were incorporated into laws, such as the Mental Deficiency Act 1913 in England. At that time, a method of classification was considered to be necessary to make possible the segregation of people whom science had deemed to be harmful to the population as a whole and a major source of criminality (Goddard, 1912). However, despite this inauspicious past, there have been substantial positive changes in the way society as a whole perceives and wishes to

engage with people with DID, and with this there have also been changes in almost every aspect of the lives of these individuals, with a focus on community inclusion and support. This chapter sets these changes in a historical context and seeks to integrate what have been very divergent and conflicting approaches. It also considers the 'models' that are helpful in thinking about the needs of people with DID and the various classification systems used.

Background

Understanding the causes and prevalence of any specific illness or disability requires that it can be accurately defined and identified. This is the bedrock of epidemiology and the investigation of aetiology and the underlying pathophysiology of ill health. Although such an approach works particularly well for investigating specific illnesses and has been central to the development of treatments, it fits less comfortably in the case of potentially lifelong disabilities such as DID. The term DID is not fundamentally a diagnosis, as its use implies very little understanding about cause, pathophysiology or likely prognosis. Furthermore, as described above, classification in DID has been associated with negative stereotyping and labelling has been used to justify actions such as segregation from society. The application of any system of classification is inevitably contradictory; on the one hand, it enables needs to be defined and for groups of people so 'labelled' to act together to argue for recognition (as seen through advocacy organisations) yet, on the other hand, the outcomes can be negative, such as dismissive attitudes. Over the years, this approach of defining and classifying has resulted in tensions between what are referred to as the 'biomedical' and the 'social' models of disability. In the papers advocating different perspectives, the language and concepts used and the conclusions drawn at times appear irreconcilable. However, although debate has been polarised, the value of each perspective and the need for a more nuanced understanding of the value of each have been increasingly recognised (Shakespeare, 2006). While each of us, with our very different professional backgrounds, places a different emphasis on the interplay between different conceptual views, there is a necessary coming together of these perspectives.

In addition to the biomedical and social models of disability, there is also a systemic model that guides understanding by seeking specifically to set disability in the context of what is often considerable complexity. The purpose of these conceptual and theoretical perspectives is that they provide a means for structuring our thinking about the needs of people with DID and, in turn, how we might respond to those needs. I very briefly consider each of these models below, before moving on to definitions and systems of classification that, at their best, provide the means for an informed and valid understanding of the person concerned and of the issues that have brought that person to the attention of services.

Theoretical models that may inform understanding

The biomedical model, central to all branches of medicine, is fundamentally diagnostic in its approach, addressing very specific questions in a particular context. For example, early in childhood this may be attempting to answer the questions that parents may ask, such as 'Why has my child not developed like other children? What are the chances that any future children will be similarly affected?' To answer these questions the approach is largely a diagnostic one – is the child's developmental trajectory atypical and does the child have a known neurodevelopmental disorder? In high-resource countries the focus is now generally on genetic disorders (and on the complex ethical issues that come with this), but in many parts of the world this biomedical approach has identified potentially preventable causes of disability, such as maternal iodine deficiency. In the latter case significant disability can be prevented through nutritional supplementation. Such an approach also seeks to understand and characterise the 'developmental trajectories' that people with specific neurodevelopmental syndromes are likely to follow across their life span, setting this understanding in the context of typical and atypical developmental profiles, such as those characteristic of autism spectrum conditions. As discussed later, this biomedical approach also provides a framework for identifying specific comorbid conditions, such as sensory impairments or physical or psychiatric illness, the treatments of which may bring benefits and reduce secondary disabilities.

The social model of disability is generally seen as the model that should drive our understanding of how social support should be structured and the philosophy that underpins policy and practice. The basic tenet of this model is that specific impairments (e.g. sensory, motor or intellectual) do not in themselves have to result in disability or disadvantage. Consequently, the social model requires individuals and society as a whole to address attitudinal and practical barriers to the full inclusion and participation of people with DID. By doing so, such disadvantage can be minimised and even eliminated. By viewing 'impairment' solely as a failure in 'an organ system' (as the biomedical model is seen to do), which in the case of people with DID may not be amenable to medical treatment, the concern is that the state may decide to abrogate any responsibility to help that person have a better life.

A further conceptual perspective that may be of value in orienting ourselves towards understanding and meeting the needs of people with DID is that described as the systemic model. It places emphasis on the idea that human behaviour should be considered and is best understood in the context of people's lives being part of a complex system. People with DID may be dependent on others for their very survival or, at the least, for enabling them to have a good life. From a health perspective, for people with DID, assessments and interventions are frequently delivered at the interface with social care. Family members or those paid to provide support are necessarily the intermediaries between those working in healthcare,

such as the general practitioner (GP), and the individuals themselves. In addition, perspectives such as that of applied behavioural analysis, which help elucidate the factors that predispose to, precipitate or maintain challenging behaviour, are very much concerned with the 'complex system' that is social care, in terms of how particular behaviours are shaped and maintained (I will return to this later in the chapter). Thus, the needs of people with DID and how best to understand and meet such needs cannot be readily separated from the wider network that surrounds each individual.

Historical context

An increasingly rapid rise in new medical and scientific knowledge took place during the 19th and 20th centuries. For example, Pasteur put forward the germ theory of disease and developed the first vaccines in the late 19th century and, in the early 20th century, Garrod proposed the concept of 'chemical individuality', recognising that individuals are importantly different in their make-up. He described alkaptonuria, an inborn autosomal recessive error of metabolism, and in doing so brought together the emerging science of genetics, as pioneered by Mendel in the 19th century, with that of chemistry (Prasad & Galbraith, 2005). Subsequently, this work on 'inborn errors of metabolism' led to the description by Følling in 1934 of phenylketonuria, an example of a rare biological cause of DID, the consequences of which are largely preventable, provided that there is early diagnosis and appropriate dietary treatment.

The accelerating nature of scientific advances is very well illustrated in the field of genetics. The structure of DNA was elucidated by Watson and Crick in 1953, and in 1956 the normal human complement of 23 pairs of chromosomes was established (Tjio & Levan, 1956). Three years later Lejeune and colleagues identified trisomy 21 as the cause of Down syndrome (Lejeune *et al*, 1959). Technologies were subsequently developed for identifying genetic variants (polymorphisms) at particular loci in the genome, which in turn led to genetic linkage studies, which were prominent in general psychiatric research by the end of the 20th century. In the year 2000, the first draft of the sequencing of the human genome was announced (International Human Genome Sequencing Consortium, 2001). DNA sequencing is now becoming increasingly possible. Over this period, the chromosomal and molecular genetic basis for many neurodevelopmental syndromes associated with DID were described. Most recently, the emphasis in genetics has moved to the expanding field of epigenetics and to an understanding of the mechanisms that regulate gene expression and the potential mechanisms whereby environmental and biological factors interact (Jirtle & Skinner, 2007).

The early 20th century saw the development of tests of intelligence. These IQ tests were initially developed as a means of distinguishing those whom we now see as having an intellectual disability from those with

mental illness, as well as for screening particular populations, such as people enlisting in the armed forces. The importance of these tests was that they provided a standardised way of comparing particular abilities and individuals' levels of performance. However, as is well known, their use was not without controversy, as IQ tests were adopted by the eugenics movement and were also used to argue the case for the belief in the inferiority of certain races (Hermstein & Murray, 1994). Wechsler tests eventually became the most established. They were standardised to a median of 100 with a standard deviation of 15 points (Wechsler, 1997). The score for any individual could then be ranked against the standardised sample for that age. Subsequent twin and family studies suggest that IQ is under considerable genetic influence and, like height, many genes may each have a small effect, a fact that is relevant when considering the aetiology of mild intellectual disability (Davis *et al*, 2010; but as a challenge to this interpretation see also an earlier paper by Devlin *et al*, 1997). Although IQ has some ability to predict an individual's level of functioning, education and opportunity are clearly of paramount importance, and perhaps the most significant advance in the 20th century for people with DID was the requirement placed on education authorities in many countries to provide education for them. Thus, as so often occurs in this area of study, developments such as the IQ test were contentious, but psychometrics in its increasingly sophisticated forms remains an important tool.

In the later 20th century there was a backlash against the over-classification and over-medicalisation of DID, particularly given the application of dubious scientific ideas to the detriment of those to whom they were applied. The horror of the consequences of misunderstanding and misapplication of scientific theory was most vividly demonstrated by the treatment of people with mental disorders, of whatever form, in Nazi Germany and afterwards by programmes of forced sterilisation.

However, despite an extremely problematic start, the 20th century was to end in very significant improvements in the lives of people with DID and in the conceptual frameworks that informed thinking, through the development of a social model of disability and an increased emphasis on human rights. Among the most striking was the changing philosophy initiated by the normalisation movement, away from segregation and towards integration and social inclusion, initially put forward by Nirje and developed by Wolfensberger (Wolfensberger, 1972). There has also been the recognition that people with DID represent a highly complex and heterogeneous group with a varied range of needs, together with an appreciation that special education, skills and communication training, and appropriate social support, can lead to levels of independence and a quality of life that were never aspired to or attained in the large institutional settings that had predominated in the care of such individuals. In the UK the scandal of abuse uncovered at Ely Hospital in Cardiff, Wales, led to the government White Paper *Better Services for the Mentally Handicapped*

(Department of Health and Social Security, 1971) and the start of the closure of long-stay institutions, once established as 'colonies' and later reborn as hospitals with the start of the National Health Service (NHS) in 1948. To the credit of successive UK governments, this enlightened and progressive approach continued with the publication of *Valuing People* in 2001 (Department of Health, 2001). In the 21st century, this fundamentally rights-based approach was established internationally in 2006 through the United Nations Convention on the Rights of Persons with Disabilities, which has now been ratified by the majority of countries across the world.

The present day

The background to developments in the understanding of intellectual disability has therefore been problematic, with systems being devised for the purpose of classification (considered below) using labels that certainly would now cause offence. Where such systems became part of the law, they were then used to legally segregate and isolate those to whom they applied. At the same time, there was an uncomfortable juxtaposition between those propounding the social model of disability, on the one hand, and, on the other, the biomedical approach and advances that were identifying specific single major causes of DID, one outcome of which was the possibility of prenatal testing. In high-resource countries, major environmental causes such as congenital rubella were identified and have essentially been eliminated through vaccination programmes, and elsewhere in the world public health measures are tackling nutritionally determined causes. Moreover, to be able to target educational support or to address some of the concerns that a family may raise when it is clear that something is wrong with their child does require a means of describing and characterising those people being referred to, and thus definitions are necessary. In addition, the variability and potential complexity of need among children and adults with DID has been highlighted by epidemiological studies that have demonstrated high rates of secondary disability due to the presence of sensory and physical impairments, behavioural and psychiatric disorders, and/or a developmental profile indicative of autism (e.g. Rutter *et al*, 1976; Cooper *et al*, 2007). The identification of such secondary impairments and disabilities and their treatment or amelioration through a range of interventions have helped to replace a feeling of therapeutic nihilism that had perhaps previously been all too pervasive. It became apparent not only that good education and social support services were required, but also that multidisciplinary and community-based specialist health support should be available to people with DID, particularly those with challenging behaviour and/or mental health problems.

The social model of disability underpins models of support for both children and adults. The biomedical model enables the identification of

specific causes of developmental delay in children. For adults with DID, it becomes important for different reasons, as the attention moves away from identifying the cause of the person's disability towards understanding, for example, the reasons for apparent ill health or the emergence of problem behaviours. Here again, different conceptual models of understanding have arisen, from the developmental to the biomedical and the psychological. One of the most influential of these has been an approach informed by learning theory that is commonly referred to as applied behavioural analysis. Applied behavioural analysis was initially developed by Lovaas as a form of intensive behavioural modification to facilitate skills development and reduce maladaptive behaviours (for a review see Matson *et al*, 1996). The importance of this approach is the contrast with the traditional biomedical model, moving attention away from the individual towards an understanding of the interaction between the individual and the environment, and how particular behaviours are shaped and maintained through reinforcement. The methods of observation developed and the subsequent coding of behaviour in order to identify antecedents and consequences now underpin much of our present-day approach to support, particularly of children and adults with autism spectrum disorders. In turn, this has led to perspectives such as those of positive behaviour support (Allen *et al*, 2005).

What the various approaches illustrate is the complexity of this field and the need for conceptually clear thinking. The challenge is to integrate different perspectives and to be able to judge what frameworks are best applied in any particular set of circumstances. Within this complex field, with its potentially competing systems of understanding, is there a role for the process of classification using systems such as DSM-5 and ICD-11? Or are there other ways of thinking about classification that might provide a better and more productive perspective?

Classification: a cautionary tale

As argued at the beginning of this chapter, the process of defining and classifying what is meant when someone is said to have a disorder of intellectual development has been a source of debate. From a positive perspective, the central principle of any system of classification is to bring order to disparate knowledge in a manner that may then enable further advances or the instigation of interventions that research has shown to be effective. In the field of DID there is no ideal or universal system – the system of classification used depends on the reasons for its use. These reasons range from the predominately administrative to the guiding of interventions and the use or not of specific treatments; definitions may also be enshrined in law, bringing with it specific powers.

During a research project that involved tracing family members (Holland & Gosden, 1990), a form was found in a file completed many

years earlier, which enabled the detention of a person in a long-stay institution under the Mental Deficiency Act 1913. The evidence used to justify segregation included the phrase 'was simple in appearance'. This phrase demonstrated both the attitudes of the time and the dangers of ill-informed and prejudicial thinking, made worse by the power that such classification has when incorporated into law. What the completed form illustrated was that being 'simple in appearance' and also being 'unable to work out change' – using the complex monetary system the UK had at the time ('doesn't know how many pennies there are in half a crown') – were essentially sufficient for the state to incarcerate this person in an institution for many years. Her four children were taken from her and placed in different families across the UK.

The dilemma, therefore, is how to target resources to those with special needs, but also to identify those with such needs in a manner that is valid and reliable and respects individual rights in a non-discriminatory way. Any system of classification inevitably has to focus on a few specific characteristics to the potential exclusion of others, and no system can impart a truly comprehensive picture. Methods of classification have therefore inevitably changed over time in an attempt to clarify the key issues and to minimise the stigma that might be associated with any given label. Some are clearly informed by one or other of the models mentioned at the beginning of this chapter, such as the biomedical model.

Conceptually, there are difficulties, as classification systems are by their very nature categorical, yet intellectual ability is clearly dimensional and continuous. Any cut-off is defined, at least partially, on the basis of IQ, and a point below which someone might be considered to have an intellectual impairment is statistically determined (two standard deviations below the mean). In contrast, particular neurodevelopmental syndromes that may be associated with DID are categorical – you have it or you do not – but even there, such obvious categorical distinctions have begun to break down as the genetic bases for syndromes are more clearly elucidated. For example, in fragile-X syndrome there is variation in the number of repeat sequences in the FMR1 mutation, in both carrier and affected individuals and across particular groups. The accepted significance of the exact number of repeat sequences, although being informed by their predictive value, in the end requires a decision as to some cut-off (below 50, above 200, etc.) (Nolin *et al*, 2003). As the significance of chromosomal copy number variants (CNVs) of various sizes is elucidated, a similar problem is likely to arise. Thus, as with conditions such as high blood pressure or diabetes mellitus, exactly what is considered normal or typical and what is considered abnormal or atypical is a judgement based on observation and research, but without necessarily a distinct separation between one and the other. I will now examine different systems of classification and then consider the relationship between assessment and classification.

The DSM and ICD systems

As set out in the Introduction to DSM-5, the first *Diagnostic and Statistical Manual* was published in 1844, to be used for the classification of the mental disorders of patients in institutional settings (American Psychiatric Association, 2013). This led to the various iterations of the DSM, most recently DSM-5, published in 2013. The ICD-11 classification is due to be published in 2017.

Both DSM-5 and ICD-11[1] place intellectual disabilities/disorders of intellectual development (DID) within the broad framework of what are termed 'mental disorders', and in that regard both are 'biomedical' in their approach. However, in DSM-5 the structure of the classification proposed has been shaped around developmental and life-span considerations and within a cultural context that recognises the dimensional nature of psychiatric disorder and how factors in the environment in which the person lives influence whether a particular symptom has functional significance or not, thereby moving beyond simply a diagnosis. Its predecessor, DSM-IV (American Psychiatric Association, 1994), provided a framework of multi-axial diagnosis, with Axis II for personality disorders and what was then termed 'mental retardation'. This is no longer the case in DSM-5. Box 1.1 summarises the DSM-5 criteria for IDD, which in essence remain as in DSM-IV, although differently worded. IDD is included in a section headed 'Neurodevelopmental disorders', which also includes communication disorders, autism spectrum disorder (without distinguishing between Asperger syndrome and autism), attention-deficit hyperactivity disorder (ADHD), specific learning disorders, motor disorders and 'other neurodevelopmental disorders'. The focus is not primarily on aetiology but rather on quantifying the extent of disability, by defining the level of intellectual impairment and listing the range of adaptive functions that might be impaired. The definition makes it explicit that the onset is in the developmental period and that IDD is the final common pathway of a number of potential aetiologies. Significant subaverage intellectual function is defined as an IQ of 70 or below (using standard IQ tests). IQ is also used to help determine the level of intellectual disability (mild, moderate, severe or profound). Adaptive functioning has to be measured against what would be expected for a person of that age, and the social and cultural experience of the person has to be taken into account. The Wechsler Intelligence Scales (to establish IQ) and the Vineland Adaptive Behavior Scales or the American Association on Intellectual and Developmental Disabilities revised Adaptive Behavior Scale (for characterising functioning) are established instruments for the measurements of these abilities and for which there are normative data for comparison.

1. It is likely that ICD-11, currently under preparation, will use the term 'disorders of intellectual development', within the category of 'Mental and behavioural disorders', but the broad framework will remain the same (http://apps.who.int/classifications/icd11/browse/l-m/en).

Box 1.1 Summary of the DSM diagnostic criteria for intellectual disabilities

- Onset during the developmental period
- Deficits in conceptual, social and practical domains
- Deficits in intellectual functions on both clinical assessment and intelligence testing (the choice of testing instrument should take into account the individual's socioeconomic background, native language and other associated handicaps)
- Deficits in adaptive functioning – how effectively individuals cope with common life demands and how well they meet the standards of personal independence expected of someone in the particular age group, sociocultural background and community setting

The degree of severity of mental retardation may be specified on the basis of intellectual impairment, taking into account other aspects of functioning.

- Mild mental retardation: IQ level 50–55 to approximately 70
- Moderate mental retardation: IQ level 35–40 to 50–55
- Severe mental retardation: IQ level 20–25 to 35–40
- Profound mental retardation: IQ level below 20 or 25

The inclusion of definitions of IDD in a manual designed to inform 'diagnosis', however, has its problems. Even where any categorisation is subdivided according to severity, it tells us very little about the cause, nor does it significantly help with intervention. Its value is to bring consistency and a degree of rigour to the classification process. Thus, it can reasonably be assumed that when properly used there will be a degree of reliability to the conclusion that someone has an IDD. It will not simply be based on appearance or educational abilities (as described earlier), but rather take into account evidence for a delayed and atypical pattern of development and the continuing presence of intellectual and functional impairments. Depending on circumstances, the next question might well be whether there is a single major cause for the developmental delay (genetic or environmental) or whether that is unlikely and a combination of factors have contributed to a person's atypical developmental history. However, in many ways, the limitations of such an approach are readily exposed, and for this reason other conceptual models have been proposed.

International Classification of Impairments, Disabilities and Handicaps

In 1980 the World Health Organization proposed a system of classification that attempted to overcome the limitations of other systems and, most

importantly, aimed to guide intervention (World Health Organization, 1980). Box 1.2 summarises the terms. In this system DID can be conceptualised at different levels. In the case of *impairment*, the organ system involved is the central nervous system. It is impairment of this system for genetic, chromosomal or environmental reasons that has primarily affected the acquisition of developmentally determined skills and the ability to learn.

Box 1.2 Definitions of impairment, disability and handicap

The *International Classification of Impairments, Disabilities and Handicaps* (World Health Organization, 1980) uses the following definitions:

Impairment

- Is any loss or abnormality of psychological, physiological or anatomical structure or function
- Represents deviation from some norm in the individual's biomedical status
- Is characterised by losses or abnormalities that may be temporary or permanent
- Includes the existence or occurrence of an anomaly, defect or loss in a limb, organ, tissue or other structure of the body, or a defect in a functional system or mechanism of the body, including the systems of mental functioning
- Is not contingent upon aetiology

Disability

- Is any restriction or lack (resulting from impairment) of ability to perform an activity in the manner or within the range considered normal for a human being
- Is concerned with compound or integrated activities expected of the person or of the body as a whole, such as represented by tasks, skills and behaviours
- Is the excesses or deficiencies of customarily expected activities and behaviour, which may be temporary or permanent, reversible or irreversible, and progressive or regressive
- Is the process through which a functional limitation expresses itself as a reality in everyday life

Handicap

- Is a disadvantage for a given individual, resulting from an impairment or disability that limits or prevents the fulfilment of a role that is normal for that individual
- Places some value upon this departure from a structural, functional or performance norm by the individual or his or her peers in the context of their culture
- Is relative to other people and represents discordance between the individual's performance or status and the expectations of his or her social/cultural group
- Is a social phenomenon, representing the social and environmental consequences for the individual stemming from his or her impairment and disability

Certainly, a key task is to identify the reasons for any abnormality of brain development and therefore intellectual impairment, and this is best done as early in the person's life as possible. It may have treatment implications, may guide prognosis and, most important, may help the parents of those affected to make sense of the disability. It may also have important implications for genetic counselling.

The associated *disability* is the effect of the impairment on a person's ability to learn and to acquire new skills that come with development. These in turn enable the acquisition of increasingly advanced skills necessary for an independent life. The exact nature and extent of the disability may include not only intellectual disabilities but also physical and sensory disabilities. The extent to which a given impairment results in a loss of function (disability) may well be influenced by the extent and nature of interventions such as special education or the correction of hearing loss by means of a hearing aid.

The final level, that of *handicap*, is a result of an interaction between the disability and the extent to which support is available or environmental adjustments are made. It is a measure of disadvantage that can be ameliorated through, for example, the provision of support or environmental modifications (e.g. wheelchair ramps) that diminish the impact of physical disabilities. Such interventions maximise independence and thereby reduce disadvantage by ensuring that the impact of any given disability on an individual's independence and quality of life is minimised. Shakespeare (2006) has argued that such a structure helps to bring together the biomedical and the social models of disability.

International Classification of Functioning, Disability and Health (ICF)

The WHO introduced this new system of classification (World Health Organization, 2001) to replace the above-mentioned International Classification of Impairments, Disabilities and Handicaps. The focus switched from a system that had been seen as just characterising the negative to a system that also emphasised the positive – what people are able to do, rather than just what they cannot do. The ICF was developed to enable the characterisation of 'health domains' across the whole population and therefore has universal application, which includes people with DID. The ICF organises information in two parts. Part 1 ('Functioning and disability') is the means whereby body functions and structures and activities and participation can be characterised. Part 2 relates to 'Contextual factors', whether in the environment or pertaining to the individual. Fig. 1.1 illustrates the relationship between the different components of this system. The WHO emphasises that it enables a multiperspective approach and, for this reason, that it provides 'the building blocks for users who wish to create models and study different aspects of

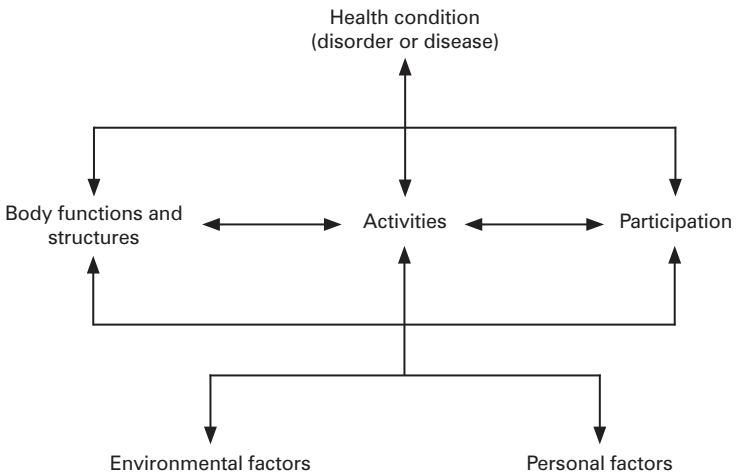

Fig. 1.1 Schematic representation of the structure of the ICF (World Health Organization, 2001: p. 18). This provides a framework for comprehensive data collection about an individual or a group of people so that needs can be characterised and comparisons made.

this process' (World Health Organization, 2001: p. 18). Thus, different disciplines can use it creatively to link to their specific scientific orientation.

Part 1 is divided into organ systems. With respect to people with DID the sections on mental functions and structures of the nervous system may be of particular relevance, but problems in these areas may be compounded by secondary disabilities, such as sensory impairments, consequent on abnormalities in the structure and function of other organ systems. All of these categories are extensively subdivided. In Part 2 the focus is very much on the specific personal circumstances and the characteristics of the individual's environment, including the health system and support available.

This system seeks to do two things. First, it aims to provide a reliable structure for the description of the complex effects of ill health and those factors that might moderate its impact, thereby enabling accurate comparisons across countries, between cultures and throughout the life span. Second, it aims to provide a more comprehensive framework to aid intervention, which moves beyond the single word or brief phrase of classification systems such as ICD and DSM, to a more structured and meaningful description of an individual's strengths and difficulties, in order to aid intervention. In this respect, it seeks to incorporate aspects of the biomedical and social models of disability and also to take a more systemic perspective, setting the person within the context of his or her support network and culture.

The prevalence and aetiology of DID

As argued, the above classification systems are not truly diagnostic. In addition, these different ways of conceptualising DID indicate that the path to functional impairments is not simply to do with biology; there are interactions with respect to the extent and nature of the statutory provision of education, opportunities in adult life and societal attitudes, all of which have an impact on ability and quality of life. The biomedical perspective is fundamentally about understanding the reasons for problems at birth or in childhood and in this regard there have been significant advances. It has been recognised for some time that there are fundamentally two broadly distinct groups, identified through population-based studies such as the Aberdeen children's cohort study of the 1950s (Birch *et al*, 1970). First, there are children who have a definite or likely genetic abnormality or specific environmental causes with major effects on subsequent development. Second, there are those in whom there is no obvious single major cause for their early developmental delay and subsequent intellectual disabilities. Here, the small effects of many genes combined with social disadvantage may be crucial. The broad differences between these two groups are summarised in Box 1.3. This 'two-group' perspective also illustrates the difficulty in arriving at a true prevalence of DID in any given community, given the fact that more than an IQ below 70 is required when determining

Box 1.3 Differences between biologically determined and subcultural DID

Biological

- Moderate/severe impairment
- Significant impairment in adaptive functioning
- Equal distribution across families of different socioeconomic status
- Parents and siblings usually of normal intelligence
- Dysmorphic characteristics common
- Other impairments and disabilities common
- Neglect unusual

Subcultural

- Mild or borderline impairment
- Minor or no impairment in adaptive functioning
- More common in families of lower socioeconomic status
- Intellectual ability impaired in family members
- Dysmorphic characteristics unlikely
- Other impairments and disabilities unusual
- Neglect more common

that someone should be considered to have a DID. The distribution of IQ in the general population is near normal, but with a skew to the left from the presence of neurodevelopmental disorders, which are generally associated with a downward shift in IQ. Given this, a figure of 2% to 2.5% is estimated as the proportion of the population with an IQ below 70. The problem then is to know how many of this group truly should be considered to have DID. To address this question there have been studies of the 'administrative prevalence' of people with DID – that is, the percentage of any given geographical population known to DID services. Figures of between 0.4% and just under 1% are arrived at, depending on a number of factors, including: whether active screening for people with DID in the population was attempted; whether it was just those people already known to DID services; whether children and adults were included; and specific geographical factors, such as levels of deprivation. Studies indicate that there is a peak in prevalence in childhood (Fryers, 2000), and that there may be significant regional variations in any given country. Examples of studies from different countries include: Sondenaa *et al* (2010) in Norway; McConkey *et al* (2006) in the island of Ireland; the European Intellectual Disability Research Network (2003) in Spain; Larson *et al* (2001) in the USA; and Wen (1997) in Australia. In an important study in the USA, Fujiura & Taylor (2003) estimated that there were a further 1.27% of people with mild intellectual disabilities who had very substantial needs and were effectively falling through the net of DID services.

The striking feature about this population is its heterogeneity, in terms of the nature and severity of disability but also of the presence or not of individuals whose DID is due to one of many possible single major causes. Making up this population there are those with conditions resulting from specific environmental factors, such as fetal exposure to alcohol, very low birth weight, congenital infections and maternal iodine deficiency (which, worldwide, is of major significance – see Zimmerman, 2009), and those with chromosomal and single-gene disorders that arise either *de novo* or are inherited. Down syndrome, due to trisomy 21, and fragile-X syndrome, due to X-linked FMR1 mutations, are two relatively common examples of the latter. Other neurodevelopmental syndromes due to copy number variants have been identified (Kaminsky *et al*, 2011) and advances in genetics using microarray technologies (sometimes referred to as molecular karyotyping) readily enable the identification of copy number variants of various sizes, including chromosomal rearrangements, deletions and duplications (Miller *et al*, 2010). There are some well-recognised deletion syndromes (e.g. Williams syndrome and *cri du chat* syndrome), but some previously unrecognised chromosomal abnormalities are still to be properly phenotypically characterised. Whether such copy number variants should be considered to be pathogenic or a normal polymorphic variation has to be judged using established databases such as DECIPHER (Firth *et al*, 2009). With improvements in DNA sequencing technology, the identification of

specific gene mutations is now more readily available. These technologies together make possible the characterisation and subsequent identification of an increasing number of neurodevelopmental syndromes of genetic origin.

Why does this matter to psychiatric practice? People with some of these syndromes have been shown to have specific developmental profiles and to be at high risk of developing certain comorbid conditions. Well known examples include the excessive eating behaviour and risk of psychopathology associated with Prader–Willi syndrome (Holland *et al*, 2003), the high rates of Alzheimer's disease affecting people with Down syndrome (Holland *et al*, 1998), anxiety disorders affecting people with Williams syndrome (Woodruff-Borden *et al*, 2010) and severe self-injurious behaviour in people with Lesch–Nyhan and Smith–Magenis syndromes (Arron *et al*, 2011). These observations, famously described by Nyhan as the 'behavioural phenotype of organic genetic disease' (Nyhan, 1971), have very significantly altered our understanding of the aetiology and pathophysiology of such behaviours and psychiatric disorders. Conceptually, comparative studies across these neurodevelopmental syndromes have challenged the orthodoxy of applied behavioural analysis; more complex models are needed, recognising the role of the syndrome-specific developmental profiles and brain mechanisms in the aetiology of syndrome-specific behaviours – see for example Karmiloff-Smith & Thomas (2003) on Williams syndrome and Reiss & Dant (2003) on fragile-X syndrome. Oliver and colleagues have, for instance, reported very different behavioural and developmental profiles across different neurodevelopmental syndromes, and they propose specific models to account for the occurrence and maintenance of specific behaviours in particular syndromes (Oliver *et al*, 2013). Holland *et al* (2003) proposed different mechanisms to account for the various components of the behavioural phenotype in Prader–Willi syndrome. High rates of psychotic illness developing in early adult life were found in the those with the syndrome, although the particularly high rates were limited to those with the chromosome 15 maternal uniparental disomy (UPD) subtype, indicating that it is not simply having Prader–Willi syndrome that increases the risk; rather, it is something unique to having a chromosome 15 maternal UPD that accounts for the increase (Boer *et al*, 2002).

The challenge is to integrate the expanding knowledge base, so as to make both conceptual and practical advances that bring real benefits to people with DID. For mental health professionals working with people with DID the focus is on the prevention and treatment of secondary disabilities that may arise in the form of maladaptive behaviours or mental ill health. Research in this area has moved from the descriptive to the epidemiological and, as indicated above, is now focusing on understanding mechanisms. Studies addressing mechanisms have been diverse and have included detailed observational clinical studies and the construction and study of genetic knockout mouse models of neurodevelopmental syndromes. The significance of this latter approach was recognised by the award of the

Nobel Prize to Mario Capecchi, Martin Evans and Oliver Smithies in 2007. The message fundamentally from these very diverse areas of research is that we must pay attention to both biology and environment when we are seeking to understand the factors that give rise to, and maintain, different constellations of problem behaviours and psychiatric disorders as they affect people with DID. Our approach to the understanding of maladaptive behaviours therefore draws from diverse perspectives including: the developmental, whereby such behaviours (e.g. repetitive behaviours) are seen as a direct consequence of a delayed or atypical developmental trajectory and may be syndrome specific (behavioural phenotype); applied behavioural analysis; and the possibility that such problems may be secondary to comorbid physical or psychiatric illness. Assessments should draw on these different perspectives and seek to arrive at an understanding in the context of the individual's life experiences and their emotional and physical environment.

The integration of perspectives

Given the complexity and heterogeneity of DID, it is clear that attempts to describe and categorise, although having their place, also have very significant limitations and, when incorporated into law, unless very carefully framed, may be misused. Tensions exist between wishing to respect an individual's autonomy and wishing to protect from harm a person seen as vulnerable. These tensions are universal. Stigma, lack of resources and punitive laws, policies and practices prevail to varying degrees in different countries and societies. Particular forms of guardianship legislation and the labelling that such laws require are still the means in some countries of enabling the lifelong segregation of individuals in large institutional settings. It is for these reasons, and because difficult judgements may have to be made, that an approach is required that can articulate and make explicit such tensions. Despite the complex ethical challenges, and through accepting the core principles that underpin approaches to community support within a social model, there have been considerable advances in our understanding of the nature and extent to which comorbid physical and mental health problems exist and how maladaptive behaviours may arise and are maintained.

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