INTRODUCTION

Epidemiology has been defined as ‘the study of the distribution and determinants of health, disease, and disorder in human populations’ (Fryers 1993). Although intellectual disability can be argued to be neither a disease nor a disorder, understanding the epidemiology of intellectual disability is of fundamental importance for service planning. Quite simply, to provide a needs-led service you have to know how many people with intellectual disabilities there are, what services they are likely to need, and whether there will be any changes in the need for services in the future.

However, determining the epidemiology and causes of intellectual disabilities is at best an inexact science. As ‘intellectual disability’ is socially constructed, what it means, how it is measured, and therefore who counts as having an ‘intellectual disability’ has varied over time (Trent 1995; Wright and Digby 1996) and across cultures and countries (Emerson et al. 2007; Jenkins 1998). Current professionally driven conceptualisations of ‘intellectual disability’ as largely a deficit in intelligence (Wright and Digby 1996) often have little resonance for people labelled with intellectual disability or their families (Finlay and Lyons 2005; Jenkins 1998). Therefore, before looking more closely at the literature concerning epidemiology and causes, we must first look at how people are currently classified as having an ‘intellectual disability’.
CLASSIFICATION

As mentioned above, ‘intellectual disability’ is socially constructed. The classification system used will determine who counts as having an ‘intellectual disability’, with obvious consequences when considering epidemiology and causes. In high-income English speaking countries, over the last 100 years classification systems have largely located intellectual disability as a series of deficits within the individual; typically in terms of deficits in intelligence and ‘adaptive behaviour’ (the behaviours necessary to function within society) that become apparent before cultural norms of adulthood (Emerson et al. 2007) – the so-called ‘medical model’. In more recent times, the social model of disability (where it isn’t a person’s ‘impairment’ that disables them, but the oppressive organisation of society that acts to create disability) has presented a fundamental challenge to traditional classification systems (Thomas 2007).

Classification systems have changed in different ways to meet the challenge laid down by the social model of disability. For example, the American Association on Mental Retardation (AAMR), now renamed the American Association on Intellectual and Developmental Disabilities (AAIDD), produced the most recent revision of their classification system in 2010 (AAIDD 2010), presented in Box 1.1; similar (although less precise) definitions are used by the Department of Health (Department of Health 2001). This revision still locates intellectual disability as largely a function of individual deficits, although in their guidance they do state that adaptive skills are a result of the ‘fit’ between a person’s capacities and their environment. In a supportive environment a person may be able to function perfectly well (thus not meeting the criteria for intellectual disability) – in a less supportive environment the same person may have problems and meet criteria for intellectual disability.

A more thoroughgoing attempt to incorporate social model ideas into medical model classification systems has come from the World Health Organization’s International Classification of Functioning, Disability and Health (ICF) (World Health Organization 2001). This classification system attempts to describe intellectual disability in terms of interactions between the person’s impairment (i.e. intellectual ability), their potential capacity and their actual performance across a range of activities, taking into account the person’s environmental, cultural and personal context.

Whichever classification system is used, there are a number of issues regarding classification which are likely to arise when working in services for people with intellectual disabilities.
Box 1.1 AAIDD 2010 Definition of ‘Intellectual Disability’

‘Intellectual disability is characterized by significant limitations both in intellectual functioning and in adaptive behaviour as expressed in conceptual, social, and practical adaptive skills. This disability originates before age 18.’

OPERATIONAL DEFINITIONS:

‘Intellectual functioning: an IQ score that is approximately two standard deviations below the mean, considering the standard error of measurement for the specific assessment instruments used and the instruments’ strengths and limitations.’

‘Adaptive behavior: performance on a standardized measure of adaptive behavior that is normed on the general population including people with and without ID that is approximately two standard deviations below the mean of either (a) one of the following three types of adaptive behavior; conceptual, social, and practical or (b) an overall score on a standardized measure of conceptual, social, and practical skills.’

Important elements of the definition:

...significant limitations ...

Intellectual disability is defined as a fundamental difficulty in learning and performing certain daily life skills. There must be significant limitations in conceptual, social and practical adaptive skills, which are specifically affected. Other areas (e.g. health, temperament) may not be.

...in intellectual functioning...

This is defined as an IQ standard score of approximately 70 to 75 or below (approximately two standard deviations below the mean), based on assessment that includes one or more individually administered general intelligence tests.

...and in adaptive behavior...

Intellectual functioning alone is insufficient to classify someone as having an intellectual disability. In addition, there must be significant
Levels of Intellectual Disability

Although some classification systems do not define levels of intellectual disability and regard the labels attached to levels of intellectual disability as misleading (AAIDD 2010), the concept of different degrees of severity of intellectual disability is commonly used in policy and practice in the UK. These classifications are typically based on standardised IQ scores. A typical system is that of the International Classification of Diseases (or ICD), produced by the World Health Organisation:

- Mild: 50–70
- Moderate: 35–49
- Severe: 20–34
- Profound: <20

For many purposes (such as epidemiological studies), all people with IQ<50 are classified as people with severe intellectual disabilities. While these labels of levels may assist heuristically in getting a sense of a person’s likely capabilities and support needs, they do not map reliably on to capabilities that are potentially important for the clinician, such as capacity to give informed consent or capacity to participate effectively in clinical interventions requiring significant linguistic, memory or other cognitive capabilities. There is no substitute for individual assessment of a person’s individual profile of capabilities and support needs.

Cultural and Linguistic Diversity

‘Intellectual disability’ is socially constructed, and can be regarded as a product of specific English-speaking cultures at a particular point in history (Emerson et al. 2007). This is particularly important when considering the reliance of epidemiological research on IQ tests, which can dramatically...
over-estimate prevalence rates of intellectual disability amongst minority ethnic communities (Hatton 2002; Leonard and Wen 2002). There are also highly likely to be cultural differences in perceptions of which behaviours are considered to be adaptive (Jenkins 1998).

Present Functioning

‘Intellectual disability’ is not necessarily a life-long trait or condition, and depending on people’s circumstances and responses to them they may not be regarded as having intellectual disabilities throughout their lives. Indeed, many people with ‘mild’ intellectual disabilities (but see AAIDD 2010) have only intermittent and time-limited contact with services, usually to assist at times of crisis.

Classification in Service Settings

Formal classification systems like the ones outlined above, with their associated assessment tools, are rarely used in existing services to make decisions about whether a person has intellectual disabilities. Also, because such assessments are made by professionals within services, decisions about whether a person has intellectual disabilities are frequently influenced by the availability of services and the professional’s judgement of what is in the best interests of the individual. Many factors can impact upon this decision; financial, political, ideological, and administrative.

Consequently, there may be people within intellectual disability services who would not meet systematic classification criteria (e.g. people who were institutionalised many years ago). It is also highly likely that there are people not in contact with intellectual disability services who would meet standard classification criteria. Services are increasingly tightening eligibility criteria to decide who is eligible for intellectual disability services and to ‘prioritise’ (i.e. ration) service provision. These eligibility criteria vary widely between different services, and use widely different methods of assessment.

EPIDEMIOLOGY

The general epidemiological literature generally has two ways of counting the number of people with a particular disorder in a given population, prevalence and incidence (see Box 1.2), although as the above discussion will have made clear this is a very inexact science when applied to people with intellectual disabilities.
Prevalence

Epidemiological studies of the prevalence of intellectual disabilities of children and adults across the world’s high income countries are becoming more common (see Leonard and Wen 2002; McLaren and Bryson 1987; Murphy et al. 1998; Roeleveld et al. 1997 for reviews). Prevalence estimates for the world’s middle and low income countries are more sparse and varied for a number of reasons, but rates may be higher than those found in high income countries (Durkin et al. 2006; Emerson and Hatton 2007; Maulik et al. 2011). Much of the variance in prevalence rates reported across studies can be accounted for by methodological factors, including:

1) Sampling method. Studies which use total population samples, and assess all members of a population for intellectual disability, typically report much higher overall prevalence rates than studies using administratively defined populations (i.e. those currently using services for people with intellectual disabilities or those known to services). This discrepancy is much less for studies of the prevalence of severe intellectual disabilities.

2) Classification criteria. As discussed earlier, classification systems for deciding whether a person has an intellectual disability vary over time and across different geographical areas, and different researchers have used more or less stringent criteria for classifying people with intellectual disabilities.

3) Assessment method. Reliance on IQ alone almost inevitably results in higher prevalence rates than those using IQ and adaptive behaviour assessment methods. Other factors, such as the skills of the professional conducting the assessment and the language and culture of people being tested (and those doing the testing) will all influence the prevalence rate reported.

Box 1.2 Definitions of prevalence and incidence

Prevalence is the number of cases, old and new, existing in a population at a given point in time or over a specified period. Incidence refers to the number of new cases of a disorder arising in a population in a stated period of time.

(Richardson and Koller 1985)
People with Mild Intellectual Disabilities (i.e. IQ 50 or 55 to 70)

Studies of high income countries (see Leonard and Wen 2002; McLaren and Bryson 1987; Murphy et al. 1998; Roeleveld et al. 1997) report the following findings (see Durkin et al. 2006, for information on low and middle income countries):

1) General prevalence rates (i.e. across all ages) of mild intellectual disabilities from 3.7 to 5.9 per 1,000 based on administratively defined populations (i.e. those known to services), with total population studies reporting much higher prevalence rates (based on IQ assessment only) of around 30 per 1,000, although a recent UK study has produced an estimated prevalence rate of 80 per 1,000 (Simonoff et al. 2006).
2) More males with mild intellectual disabilities than females (ratio approx. 1.6:1, although gender ratios vary widely across studies).
3) An increase in the apparent prevalence of mild intellectual disabilities throughout the school years, followed by a sharp drop around the school leaving age.
4) A disproportionate number of people with mild intellectual disabilities come from disadvantaged socio-economic backgrounds.

These findings illustrate clearly the complex processes involved in the classification of people as having intellectual disabilities. For example, changes in age-specific prevalence rates (e.g. increasing across school age, then dropping beyond school-leaving age) may simply reflect people’s identification by and ongoing contact with services, or they may be a consequence of people on adulthood demonstrating a good adaptive fit to their circumstances, thereby no longer meeting the classification criteria for intellectual disability.

People With Severe Intellectual Disabilities (i.e. IQ < 50 or 55)

Studies of high income countries (see Leonard and Wen 2002; McLaren and Bryson 1987; Murphy et al. 1998; Roeleveld et al. 1997) report the following findings (see Durkin et al. 2006, for information on low and middle income countries, where prevalence rates are likely to be at least double):

1) General prevalence rates (i.e. across all ages) of severe intellectual disabilities from 3 to 4 per 1,000, with total population studies reporting higher prevalence rates (e.g. 6.3 per 1,000) than studies using administratively defined populations.
2) More males with severe intellectual disabilities than females (ratio approx 1.2:1).
3) An increase in the apparent prevalence of severe intellectual disabilities throughout the school years, with little if any reduction at school-leaving age.
4) Less association between prevalence and socio-economic background.

For people with severe intellectual disabilities, the classification of intellectual disability is usually made with more confidence and earlier than for people with mild intellectual disabilities. Also, children with severe intellectual disabilities are likely to continue receiving services after leaving school, and are less likely to move out of a classification of intellectual disability due to changes in circumstances.

**Incidence**

Epidemiological studies of the incidence of intellectual disability are scarcer than studies of prevalence, largely due to their methodological difficulty (Fryers 1993). These studies tend to rely on administratively defined populations of people with intellectual disabilities (typically people identified as such by service systems) rather than independently assessing entire populations, resulting in possible underestimates of incidence for people with mild intellectual disabilities. Studies across the US and Northern Europe have reported similar incidence rates – for example a US study reported a cumulative incidence at age 8 years of 4.9 children with severe intellectual disabilities per 1,000 births and 4.3 children with mild intellectual disabilities per 1,000 births (Katusic et al. 1995; see also Rantakallio and von Wendt 1986).

**UK Trends – Prevalence and Service Need**

It is not possible to estimate the number of number of people with intellectual disabilities in the UK either from information held by central government departments or from large-scale population based surveys. Robust estimates of current and future numbers of people with intellectual disabilities, based on data from registers and general population data, have been recently derived (Emerson and Hatton 2008, 2011). These estimates conclude that:

- 169,000 people aged 20 or more (0.46% of the adult population) were known users of learning disability services in England. Of these, 26,000 were aged 60 or more.
985,000 people in England have an intellectual disability, including people not identified by specialist services (2% of the general population). This included 796,000 people aged 20 or more, of whom 174,000 were people aged 60 or more.

Overall, there will be sustained growth over the next two decades in both the numbers of people with intellectual disabilities known to learning disability services (11% over the decade 2001–2011, 14% over the two decades 2001–2021) and the estimated ‘true’ number of people with intellectual disabilities in England (15% over the decade 2001–2011, 20% over the two decades 2001–2021).

Within the 50+ age range there will be very marked increases in both the numbers of people with learning disabilities known to learning disability services (28% over the decade 2001–2011, 48% over the two decades 2001–2021) and in the estimated ‘true’ number of people with intellectual disabilities in England (31% over the decade 2001–2011, 53% over the two decades 2001–2021).

When considering the service needs of people with intellectual disabilities in the UK, three issues stand out:

1) In common with other high income countries, the life expectancy of people with intellectual disabilities in the UK is increasing, although still lower than that of the general population (Carter and Jancar 1983; Hollins et al. 1998; McGuigan et al. 1995). Combined with bulges in prevalence rates for the ‘baby boom’ generation (Fryers 1993) and the increasing survival throughout adulthood of people with more complex and multiple needs, this increased life expectancy suggests a sharp rise in demand for adult services, particularly housing support services.

2) There is some tentative evidence (Emerson and Hatton 2004; Emerson et al. 1997; Kerr 2001; but see McGrother et al. 2002) of higher prevalence rates of severe intellectual disability for children in some South Asian communities in the UK. Combined with the relatively young age structure of minority ethnic communities in the UK, the need for services directed to South Asian people with intellectual disabilities (and possibly other minority ethnic communities, although prevalence data are unavailable) will continue to increase over the next 20 years (Emerson and Hatton 1999).

3) There is a consensus among practitioners that increasing numbers of children with intellectual disabilities and very complex medical needs are surviving into adulthood as a result of improved medical care and nutrition (e.g. tube feeding). At present, however, no data are available with which to evaluate this claim.
Disorders and Conditions Associated with Intellectual Disabilities

A range of disorders have been found to be more likely amongst people with intellectual disabilities. While the number of additional disorders a person is likely to have increases with the severity of the intellectual disability, the type of additional disorder reported does not seem to vary significantly across the range of intellectual disabilities (McLaren and Bryson 1987). The wide variation in prevalence rates for associated disorders reflect a wide range of methodological, classification, and assessment differences across studies, and therefore should not be treated as definitive. The most common disorders or conditions associated with intellectual disabilities include:

1) **Epilepsy.** Between 15% and 30% of people with intellectual disabilities have been reported to have epilepsy (McLaren and Bryson 1987).

2) **Cerebral palsy/other motor impairments;** reported in 20% to 30% of people with intellectual disabilities (McLaren and Bryson 1987).

3) **Sensory impairments;** reported in 10% to 33% of people with intellectual disabilities, although studies using clinical criteria for sensory impairments report much higher rates than studies using functional criteria (Hatton and Emerson 1995; McLaren and Bryson 1987).

4) **Challenging behaviour;** reported in 6% to 14% of people with intellectual disabilities, although there are widely different criteria for determining and measuring challenging behaviour (Emerson 2001; McLaren and Bryson 1987).

5) **Mental health problems.** Due to difficulties in accurately identifying mental health problems in people with intellectual disabilities and differences in definition, the range of prevalence rates reported are particularly wide (10% to 80%), although studies using more stringent criteria tend to report rates of mental health problems among adults (around 20% to 40%) similar to or slightly higher than the general population (Hatton and Taylor 2005). Among children, there is growing evidence of a four to five fold elevation in rates of mental health problems among children with intellectual disabilities (Einfeld and Emerson 2008; Emerson and Hatton 2007).

**CAUSES OF INTELLECTUAL DISABILITY**

Understanding the causes of a person’s intellectual disability can have a potentially crucial impact on prevention, treatment and management programmes for that individual (AAIDD 2010). The ‘new genetics’ (Dykens et al. 2000) is driving much of the research attempting to link genetic
'causes' to treatment and management, although much of this work is the subject of intense ethical debate. For clinical psychologists, the most important issue is that understanding the cause of a person’s intellectual disability may have implications for management programmes. One well-known example is phenylketonuria, a deficit in metabolising a particular protein which causes severe intellectual disabilities if untreated. However, such intellectual disabilities can be completely avoided by a diet low in the protein phenylalanine.

The ‘new genetics’ has also introduced the concept of the ‘behavioural phenotype’ (Dykens et al. 2000; Hodapp and Dykens 2004). Prototypically, ‘a behavioral phenotype should consist of a distinct behavior that occurs in almost every case of a genetic or chromosomal disorder, and rarely (if at all) in other conditions’ (Flynt and Yule 1994, p. 666). This strict definition would limit the study of behavioural phenotypes to a small number of genetic/chromosomal conditions with clear behavioural consequences (e.g. self-mutilation in Lesch-Nyhan syndrome; hyperphagia in Prader-Willi syndrome). This original concept of behavioral phenotypes has been broadened to a more probabilistic definition, for example ‘the heightened probability or likelihood that people with a given syndrome will exhibit certain behavioral and developmental sequelae relative to those without the syndrome’ (Dykens 1995; p. 523). Clearly, understanding the behavioural phenotype associated with a specific condition is vital when supporting that individual, for example in terms of the person’s physical environment, routines, learning style, challenging behaviours, etc. (Dykens et al. 2000). However, some caution needs to be expressed regarding the behavioural phenotype approach. Correspondences between genetic syndrome and particular behaviours are rarely perfect; many people with the genetic syndrome do not show the behaviour and many people without the genetic syndrome do show the behaviour. There is also a danger that self-fulfilling prophecies may occur within services (for example, the belief that if a person has a particular genetic syndrome, then particular behaviours are inevitable and not amenable to intervention).

The incidence, prevalence and consequences of different aetiologies are dynamic over time, due to factors such as changes in the age of parents at the child’s birth, changes in parental health behaviours such as smoking and alcohol use prenatally, prenatal screening programmes, the survival and treatment of low birthweight babies, and interventions for specific factors associated with intellectual disabilities (Brosco et al. 2006; Leonard and Wen 2002), although overall prevalence rates seem relatively stable (Leonard and Wen 2002).

Generally, studies estimate that for people with severe intellectual disabilities, aetiology is unknown for between 20% and 40% of cases, although figures in recent studies are at the lower end of this range.
For people with mild intellectual disabilities, aetiology is unknown for a somewhat higher 45% to 62% of cases (McLaren and Bryson 1987). For perhaps the majority of people, the determinants of intellectual disability will involve a complex interaction between biomedical, social, behavioural and educational factors. These factors may influence the individual at the prenatal, perinatal, and postnatal stages of life.

Prenatal Causes

Overall, studies estimate that more than 20% of cases of severe intellectual disability can be accounted for by chromosomal disorders, and that a further 20% to 40% of cases accounted for by other prenatal factors, such as single gene disorders, multi-factorial/polygenetic causes and environmental effects (McLaren and Bryson 1987; Partington et al. 2000). For people with mild intellectual disabilities, only 4% to 10% of cases are generally accounted for by chromosomal disorders (McLaren and Bryson 1987; Matilainen et al. 1995), with a further 11% to 23% of cases assumed to be due to other prenatal causes (McLaren and Bryson 1987; Matilainen et al. 1995).

Biomedical Factors

Prenatal biomedical factors potentially determining intellectual disability include chromosomal disorders, single gene disorders and other syndrome disorders (see Connor and Ferguson-Smith 1993; Plomin et al. 1997). Advances in medical genetics are being made at a rapid pace, with an increasing number of genetic abnormalities and associated syndromes being identified, although how many cases of currently unknown aetiology will become newly identified is open to debate (McLaren and Bryson 1987).

Chromosomal disorders These account for between 20% and 40% of all live births of people with severe intellectual disabilities. This range of estimates possibly reflects differences across studies in the availability of amniocentesis and genetic screening, and differences in maternal age (McLaren and Bryson 1987). The majority of conceptions with chromosomal disorders spontaneously abort (Connor and Ferguson-Smith 1993).

By far the most common chromosomal disorder associated with intellectual disability is Down syndrome (Trisomy 21, where a person has an extra whole or part chromosome-21). Approximately 1 in 700 live births have Trisomy 21, and almost all people with Down syndrome have an additional intellectual disability to some degree. People with Down syndrome are at risk for congenital heart problems, thyroid problems, epilepsy, immunological deficiencies, vision and hearing loss, and reduced
life-span with early-onset dementia (Dykens et al. 2000). Other less common chromosomal disorders are listed in Table 1.1.

**Single gene disorders** In recent years, there has been increasing interest in Fragile-X syndrome, which occurs more commonly in males, and has been claimed to be the most common hereditary cause of intellectual disability. Recent estimates of the frequency of Fragile-X are at around 1 in 4000 males and at least half as many females (Dykens et al. 2000). Common (although not conclusive) indicators of Fragile-X include an elongated face with large ears, and enlarged testes in males. Other single-gene disorders associated with intellectual disabilities are listed in Table 1.1.

**Other syndrome disorders** There are a wide range of other relatively rare biomedical syndromes that may have a prenatal causal effect on later intellectual disabilities. These often have a genetic basis in that they are the result of dominant or recessive genes, but they may also have a polygenetic basis, and vary widely in the severity of the effect. These include neurofibromatosis, tuberous sclerosis, myotonic dystrophy, craniosynostosis syndromes, and inborn errors of metabolism. Other prenatal biomedical causes of intellectual disabilities may not be associated with genetic disorders, but may be the result of disorders of brain formation at the prenatal stage. Spina bifida is probably the most well known of this group of disorders (Connor and Ferguson-Smith 1993).

**Environmental Factors**

Whilst the importance of prenatal environmental effects has long been recognised, assessing the degree of their causal influence on the later development of intellectual disabilities has still to be conclusively determined. Studies have produced a wide range of estimates for the number of cases attributable to the prenatal environment (0.7% to 11.2% for people with severe intellectual disabilities; 8.2% to 8.8% for people with mild intellectual disabilities; McLaren and Bryson 1987; Matilainen et al. 1995). Factors here include maternal malnutrition and ingestion of drugs and toxins during pregnancy (e.g. fetal alcohol syndrome), maternal diseases during pregnancy, and irradiation during pregnancy, although the relative impact of these factors in influencing intellectual disability is unknown (McLaren and Bryson 1987).

**Perinatal Causes**

Overall, studies estimate that approximately 10% of cases of severe intellectual disability are due to perinatal causes. Figures for mild intellectual disability are more variable, ranging from 1% to 19% of cases (McLaren and Bryson 1987; Matilainen et al. 1995).
Table 1.1 Genetic disorders associated with intellectual disabilities (from Connor and Ferguson-Smith 1993; Einfeld and Emerson 2008; McLaren and Bryson 1987; Plomin et al. 1997).

**Chromosomal Disorders**

<table>
<thead>
<tr>
<th>Name</th>
<th>Syndrome Name</th>
<th>Birth Prevalence</th>
<th>Associated Features</th>
</tr>
</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>Down’s syndrome</td>
<td>1 in 700 (related to maternal age)</td>
<td>Almost all some intellectual disability. Most survive well into adulthood; some evidence of early onset dementia.</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>Edward’s syndrome</td>
<td>1 in 3,000</td>
<td>All have severe intellectual disability. 10% survive to age one year.</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>Patau’s syndrome</td>
<td>1 in 5,000</td>
<td>All severe intellectual disability / seizures. 18% survive to age one year.</td>
</tr>
<tr>
<td>15q- (maternal)</td>
<td>Angelman syndrome</td>
<td>1 in 20,000</td>
<td>Generally moderate intellectual disability.</td>
</tr>
<tr>
<td>15q- (paternal)</td>
<td>Prader-Willi syndrome</td>
<td>1 in 10,000</td>
<td>Almost all some intellectual disability. Short stature, over-eating.</td>
</tr>
<tr>
<td>7p-</td>
<td>Williams syndrome</td>
<td>1 in 25,000</td>
<td>Almost all some intellectual disability by later childhood.</td>
</tr>
<tr>
<td>5p-</td>
<td>Cri-du-chat syndrome</td>
<td>Very rare</td>
<td>Half show severe intellectual disability.</td>
</tr>
<tr>
<td>Klinefelter syndrome (XXY)</td>
<td></td>
<td>1 in 1,000 males</td>
<td>Some reduction in verbal skills, severe intellectual disability uncommon.</td>
</tr>
</tbody>
</table>

Other very rare chromosomal disorders include Trisomy 8, 9p+ and 4p-
<table>
<thead>
<tr>
<th>Single Gene Disorders</th>
<th>1 in 4,000 males; at least half in females</th>
<th>Mild-moderate intellectual disability in one third of boys and girls.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fragile-X syndrome (X-linked)</td>
<td>1 in 20,000 males</td>
<td>Almost all severe intellectual disability. All show severe self-injury, usually hand and lip biting.</td>
</tr>
<tr>
<td>Lesch-Nyhan syndrome (X-linked)</td>
<td>1 in 3,500 males</td>
<td>Variable effect on intellectual ability: verbal skills more impaired. Progressive muscle wasting, almost all die before age 20.</td>
</tr>
<tr>
<td>Duchenne’s muscular dystrophy</td>
<td>1 in 10,000</td>
<td>Untreated, some severe intellectual disability.</td>
</tr>
<tr>
<td>Phenylketonuria</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Biomedical Factors

Intrauterine infections are the most common biomedical perinatal cause of intellectual disabilities, accounting for 2% to 6% of people with severe intellectual disabilities, and 1% of people with mild intellectual disabilities. Such infections include cytomegalovirus, toxoplasmosis, rubella, neonatal herpes and bacterial meningitis (McLaren and Bryson 1987).

Environmental Factors

The most common perinatal cause of severe intellectual disability is asphyxia (lack of oxygen), with 4% to 8% of people with severe intellectual disability suffering asphyxia during birth. The figures for asphyxia in people with mild intellectual disabilities vary widely, from 5% to 19%. Another common cause of intellectual disability is premature birth of the child (3% to 5% of people with severe intellectual disabilities; figures for people with mild intellectual disabilities unknown). Other perinatal traumas that may result in intellectual disability include haemorrhaging in the brain due to abnormal labour and/or delivery, premature birth, and umbilical cord accidents (McLaren and Bryson 1987).

Postnatal Causes

Very little is known about the relative impact of postnatal factors on the development of intellectual disabilities. Studies of people with severe intellectual disabilities report a wide range of estimates (1% to 13%) of people influenced by postnatal factors (McLaren and Bryson 1987; Matilainen et al. 1995). For people with mild intellectual disabilities, there is a common assumption that environmental factors, particularly those associated with socio-economic disadvantage, are the predominant causal factor, although these links are usually inferred rather than demonstrated (Emerson et al. 2007; McLaren and Bryson 1987).

Biomedical Factors

There are a wide range of postnatal biomedical factors that may impact upon the development of intellectual disability, although estimates of their prevalence are as yet unavailable. Such factors include infections (e.g. encephalitis, meningitis), diseases affecting the central nervous system, degenerative disorders (e.g. Rett syndrome, Friedrich’s ataxia and basal ganglia disorders), and epilepsy and related disorders (Connor and Ferguson-Smith 1993).

Environmental Factors

Again, a wide range of environmental factors, probably in a multifactorial fashion, can influence the development of intellectual disabilities. Traumatic
injuries to the brain, various toxic disorders (e.g. high levels of lead or mercury, dehydration, hypoglycemia) and malnutrition can all cause intellectual disabilities. Environmental deprivation, such as psychosocial disadvantage, child abuse and neglect, and chronic social/sensory deprivation, have all been hypothesised to influence the development of intellectual disability, although the relative impact they have compared to other factors is largely unknown (Durkin et al. 2006; Einfeld and Emerson 2008; Emerson et al. 2007; World Health Organization and World Bank 2011).

CONCLUSIONS

Understanding the epidemiology and causes of intellectual disabilities is important for developing needs-led services, and developing treatment and management programmes for people with intellectual disabilities. As ‘intellectual disability’ is socially constructed and subject to variations in definition and identification, studying the epidemiology and causes of intellectual disability is an at best inexact science. Despite these variations, it is clear that there are substantial and increasing numbers of people with intellectual disabilities who require a variety of services. Research is developing more sophisticated methods of identifying potential causes of intellectual disability, tracking their behavioural effects, and therefore suggesting new methods for treatment and management.

FURTHER READING


References


