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786 Chapter 17 Intellectual disability Introduction Patients with an ID have unique needs, sufficiently different from those of the general population as to require specialist psychiatric services. The incidence of mental illness is 72–3 times that of the general population, and illness often presents in a different manner. It is a complex subspecialty, encompassing everything, from molecular genetic diagnostic techniques to provision of adequate social supports, and which requires an enquiring mind and a truly holistic approach to medicine. Psychiatrists in ID will be involved in the assessment and treatment of acute and chronic mental illness, challenging behaviour, and pervasive developmental disorders, and will require a more detailed knowledge of how physical illness, epilepsy, sensory impairments, and environmental factors affect the presentation of mental disorders. While challenging, it can be a very rewarding specialty where appropriate management can dramatically improve the quality of life for patients. The ID psychiatrist will work with children, adolescents, adults, the elderly, families, and carers in a variety of clinical settings, usually in collaboration with a range of other professionals in a variety of disciplines:

- Paediatricians, during childhood, but particularly at the time of establishing the cause of developmental delay.
- Clinical geneticists and genetic counsellors, either in childhood or in adulthood, when looking both at the individual and the family for potential genetic causes.
- Social work departments when setting up and reviewing appropriate packages of care, particularly when an MHA or a capacity/incapacity legislative framework is in place.
- Psychologists are often involved in both the assessment and subsequent management of patients with ID. They will often use mixed methods, drawing on behavioural, cognitive, and dynamic approaches. In some services, music therapists and art therapists are involved in individual and group work.
- Speech and language therapists play an important role in the assessment and management of people with an ID and are vital members of the multidisciplinary health team.
- Occupational therapists, physiotherapists, and dietitians all have particular roles in the ID team, more so than in general

adult psychiatry, and, as such, are often full-time members of the team. • Family/carers are at the 'coal face' of care, and it must always be remembered that they have often years of experience with their child/ward. It is essential to engage the family/carers in the assessment and management. The ID psychiatrist will often act as a focal point in the collation and dissemination of information, being a 'fixed point' for the family/carers who may be somewhat 'at sea' with the dizzying array of professionals involved in the care of the child or adult with ID.

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788 Chapter 17 Intellectual disability Historical perspective The Mental Deficiency Act (1913) and the Elementary Education (Defective and Epileptic Children) Act (1914) were turning points in the management of those diagnosed as 'mentally defective' or 'feeble-minded' (by 'duly qualified' medical practitioners) in the UK, requiring local authorities to provide suitable care in special institutions or the guardianship of families and educational placements in special schools or classes. These 'segregation' acts moved those with ID from home, asylum, or workhouse to special institutions, with the aim of providing for their special needs and the hope of social treatments (through education and training). The motivation of at least some of those who advocated institutional care may have been admirable. Unfortunately, the definition of ID (defined as 'idiots', 'imbeciles', and the 'feeble-minded') was related to subjective measures, such as the 'ability to care for oneself', rather than objective measures such as intelligence. This led to abuses such as other 'deviant behaviours' (e.g. having an illegitimate child, habitual drunkenness) being used as grounds for committal to an institution. In addition, the institutions became focuses for contemporary social concerns, by scapegoating the 'feeble-minded' as the cause of everything, from social problems (e.g. poverty, alcoholism, unemployment, promiscuity, illegitimacy) to imperial, and even racial, decline. Progress was gradually made in the use of more objective assessment of 'defectives', but most medical authorities believed causation was inherited (a 'neuropathic trait'). This fed directly into prevalent eugenic notions of preventing 'racial decline' by segregation, with physical stigmata (e.g. facial characteristics) seen as 'proof' that appearance (especially 'racial characteristics') and mental health were interrelated. Nowadays, such ideas seem simplistic (like the practice of phrenology at the time), but the notion that the Caucasian races were 'more civilized' had significant influence at the beginning of the twentieth century. Some doctors even advocated compulsory sterilization 'to protect social health, but permit liberty'. It would take decades, and two World Wars, before social, political, and scientific pressure finally dismantled these firmly held ideas. Impetus for change came from growing concerns about the effects of large institutions, the forms of treatment, and the rights of those with ID. In the 1960s, official enquiries found evidence of abuse, malpractice, and neglect. Alarm among social reformers about the conditions in institutions was fuelled by Erving Goffman's *Asylums*. Efforts were made to reduce stigma by replacing older labels with less pejorative terms (e.g. 'mental subnormality', 'mental retardation', and 'mental handicap' for 'mental deficiency'; 'idiot', 'imbecile', 'trisomy 21', or 'Down's syndrome' for 'mongolism'; 'congenital hypothyroidism' for 'cretinism'). In 1968, ICD-8 (WHO) classified 'mental retardation' according to the severity of intellectual impairment (by IQ assessment) and social factors. The 1970s and 1980s saw major policy changes, emphasizing integration with mainstream resources and education, away from institutions and to the community. Many people with ID moved from hospitals to purpose-built hostels or 'group homes'. Understanding of the aetiology of ID expanded from the 1950s onwards, with Lionel Penrose's *Biology of Mental Defect* in 1949 and the

Historical perspective discovery of the genetic basis of Down's syndrome by Jérôme Lejeune in 1959. By the 1970s, most standard textbooks recognized multiple aetiologies (genetic and environmental), separating pre-, peri-, and postnatal causes. Karyotyping, identifying metabolic abnormalities, and isolating infectious agents allowed for laboratory diagnoses, rather than reliance on clinical observation. Pharmacological treatments of epilepsy, behavioural disturbance, movement disorders, and psychiatric comorbidity; dietary treatments of metabolic disturbances; behavioural and cognitive approaches; improved assessment/management of social/occupational functioning, communication problems, and educational needs have allowed rational management of ID. The last 20yrs have seen enormous changes in the way that people with IDs are viewed and the way in which they are treated. The large institutions are largely gone, and indeed many of the small hospitals as well. The majority of patients live either in their own homes or in a small community placement with paid carers. While this has undoubtedly benefits when compared to the large, anonymous institutions, it has created an entirely new set of challenges and problems. Some patients miss the social aspects of the group setting and are frustrated that the only people with whom they have contact are paid carers/support workers. The design of care provision has come a long way, but there will always be a need to keep on improving. The clinical terms used to refer to individuals with ID have changed over the years, as formerly neutral terms have acquired pejorative connotations and been replaced. When DSM-5 was published, it used the term 'intellectual disability', rather than 'mental retardation' (see Box 17.1). ICD-11 has replaced the term 'mental retardation' with 'disorder of intellectual development' (provisional, mild, moderate, severe, profound, and unspecified), and the Royal College of Psychiatrists have renamed the previous 'Learning Disability' faculty to 'Intellectual Disability', in line with international nomenclature. In the UK, these terms all have the same meaning. Box 17.1 'Rosa's Law' (US Federal Statute, Public Law 111-256) Rosa Marcellino, an 8-year-old girl with Down's syndrome from Maryland, was taunted frequently and pejoratively called 'retard' in a demeaning manner. With support from her state representative and US Senator Barbara Mikulski, legislation was initiated, leading to the change in the law, replacing the term 'mental retardation' with 'intellectual disability'. <http://www.gpo.gov/fdsys/pkg/PLAW-111publ256/pdf/PLAW-111publ256.pdf> [accessed 11 July 2018].

790 Chapter 17 Intellectual disability Classification ICD-10 and DSM-IV previously agreed on the use of the terms mild, moderate, severe, and profound to describe the degree of ID or 'mental retardation' (ICD-10), with arbitrary cut-offs varying only slightly (see Table 17.1). DSM-5 no longer quotes IQ scores in the diagnostic criteria, although they are still included in the description of ID with an IQ of <70 (2 standard deviations below the mean), considered to be an indication of ID. The severity of ID now includes measures of both deficits in intellectual functions and adaptive functioning. The same applies to proposed ICD-11 criteria of 'significantly below average intellectual functioning and adaptive behaviour' that are 2-3 (mild), 3-4 (moderate), or >4 (severe/profound) standard deviations below the mean. In ICD-11, severe and profound disorders of intellectual development are differentiated exclusively on the basis of adaptive behaviour differences. ICD-10 guidelines ICD-10 defines 'mental retardation' as 'a condition of arrested or incomplete development of the mind, characterized by impairments of skills manifested in the developmental period, i.e. cognitive, language, motor, and social abilities'. Mild Delay in acquiring speech, but eventual ability to use everyday speech; generally able to independently self-care; main problems in academic settings (e.g. reading, writing); potentially capable of working; variable degree of emotional and social immaturity; problems more like the normal population.

Minority with a clear organic aetiology, variable associated problems (autism, developmental disorders, epilepsy, CDs, neurological and physical disabilities). Moderate Delay in acquiring speech, with ultimate deficits in use of language and comprehension; few acquire numeracy and literacy; occasionally capable of simple supervised work. Majority have an identifiable organic aetiology, and a substantial minority have associated problems (autism, developmental disorders, epilepsy, CDs, neurological and physical disabilities). Severe Similar to moderate, but with lower levels of achievement of visuospatial, language, or social skills. Marked motor impairment and associated deficits.

Table 17.1 Classification IQ range for categories ICD-10 DSM-IV Mild 50–69 50–55 to 70 Moderate 35–49 35–40 to 50–55 Severe 20–34 20–25 to 35–40 Profound Below 20 Below 20–25

Classification Profound Comprehension and use of language very limited; basic skills limited at best; organic aetiology clear in most cases; severe neurological and physical disabilities affecting mobility common; associated problems (atypical autism, pervasive developmental disorders, epilepsy, visual and hearing impairment) more common. DSM-5 criteria • Deficits in intellectual functions confirmed by both clinical judgement and individualized standardized intelligence testing (e.g. reasoning, problem-solving, planning, abstract thinking, judgement, academic learning, learning from experience). • Deficits in adaptive functioning resulting in failure to meet developmental and sociocultural standards for personal independence and social responsibility. Without support, these deficits limit functioning in one or more activities of daily life (e.g. communication, social participation, independent living) across multiple environments (e.g. home, school, work, community). • Onset as for ICD-10: ‘during the developmental period’, discriminating ID from ABI. • Guidance on the assessment of severity suggests consideration of three domains: • Conceptual: language, reading, writing, maths, reasoning, knowledge, and memory. • Social: empathy, social judgement, communication skills, ability to make and retain friendships. • Practical: personal care, job responsibilities, money management, recreation, school tasks.

‘Subcultural’ intellectual disability Although the concept of ‘psychosocial’ causation (due to physical and emotional neglect) is controversial, it is true to say that mild or borderline intellectual impairment is more common in families of lower socio-economic status. This is best viewed as a cultural norm, and individuals generally have no, or only minor, impairments in adaptive functioning (i.e. lack of disability) (E Disability, p. 792). Generally the intellectual ability of family members is also in the borderline range, dysmorphic characteristics are less likely, and other impairments or disabilities are unusual. This is in contrast to biological causation where impairments are more significant, there is no difference in socio-economic status, parents and siblings are usually of normal intelligence, and dysmorphic features are more common.

792 Chapter 17 Intellectual disability Disability Disability is an inherently difficult concept to define and depends on a complex interplay between the person and their environment. It is not a diagnostic term, and nor are the phrases ‘intellectual disability’ or ‘mental retardation’. Instead these are descriptions of impairments of functioning on various levels, which have an aetiology that is known (e.g. Down’s syndrome) or unknown (e.g. childhood disintegrative disorder). The International Classification of Functioning, Disability, and Health (ICF) First introduced by the WHO1 as a means of trying to establish a standardized approach to describing health and health-related domains. The current version was established in 2001 and is more focused on describing people’s levels of functioning, rather than on disability. It aims to establish disability as something that can occur in everyone to some degree, rather than being something stigmatizing. The ICF describes

disability and functioning as umbrella terms denoting the positive and negative aspects of functioning from a biological, individual, and social perspective. Functioning is regarded as the dynamic interaction between a person's health condition, environmental (external) factors, and personal (internal) factors, and ICF organizes this information in two parts. Part 1 deals with functioning and disability, while Part 2 covers contextual factors. There are three levels of functioning, and disability results from dysfunction of these levels:

- Body functions and structures (physiological and anatomical systems)—problems with these cause impairment.
- Activities (carrying out a task or action)—issues with this cause activity limitation.
- Participation (involvement in a situation)—issues with this cause participation restrictions.

There is a complex, dynamic, and often unpredictable relationship between these entities. Simple linear inferences are too reductionistic, as the interactions go both ways (see Fig. 17.1). Judging overall disability from a diagnosis alone and ignoring personal factors when assessing participation or environmental factors when assessing activity limitation are likely to give a false picture of an individual's functional capacity. It is vital to collect data on these various entities independently and to empirically explore the associations between them.

1 World Health Organization (2001) International Classification of Functioning, Disability, and Health (ICF). M <http://www.who.int/classifications/icf/en/> [accessed 11 July 2018].

Disability The WHO Disability Assessment Schedule The WHO Disability Assessment Schedule (WHODAS 2.0, 2010)² is an assessment tool that accompanies the ICF and gives standardized measurements of health and disability. It scores functioning in six different areas over the previous 30 days:

- Cognition—how well someone is able to understand and communicate.
- Mobility—how well someone is able to get around.
- Self-care—how well someone can manage their personal care.
- Getting along—how well someone manages to interact with others.
- Life activities—how well someone manages in domestic, leisure, and occupational settings.
- Participation—how well someone manages to join in community activities.

Once scoring is completed, this equates to a metric range of 0–100 (with zero being classed as 'no disability' and 100 being 'full disability').

International Classification of Function Model (ICF)

Health condition (disorder or disease)	Activity	Participation	Contextual factors	Personal factors	Environmental factors
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Body functions and structure

Fig. 17.1 The ICF model. Source: data from World Health Organisation (2001) International Classification of Functioning, Disability and Health (ICF). See WHO website: M <http://www.who.int/classifications/icf/en/> [accessed 11 July 2018].

2 World Health Organization (2010) Measuring health and disability: manual for WHO Disability Assessment Schedule (WHODAS 2.0) M http://apps.who.int/iris/bitstream/10665/43974/1/9789241547598_eng.pdf?ua=1 [accessed 11 July 2018].

794 Chapter 17 Intellectual disability Aetiology A specific cause for ID can be identified in about 80% of severe and 50% of mild cases. Modern classifications of aetiological factors are based on timing of the causative event (see Table 17.2)—about 50–70% of cases will be attributable to a prenatal factor, 10–20% to a perinatal factor, and 5–10% to a postnatal factor. The identification of aetiological factors is important, because it allows for discussion of the risk of recurrence in future pregnancies. A known cause can allow for discussion of likely disabilities, possible cognitive impairments, and prognosis. This can be useful for planning supports/services, access to education, and optimizing environmental factors.

Genetic causes

- Autosomal chromosome disorders (e.g. Down's syndrome; E Down's syndrome, p. 806).
- Sex chromosome disorders (E Sex chromosome disorders, p. 817).
- Deletions and duplications (E Deletions and duplication syndromes, p. 808).

Autosomal dominant (E Autosomal dominant syndromes, p. 810) and recessive (E Autosomal recessive syndromes, p. 812) conditions. • X-linked recessive (E X-linked recessive syndromes, p. 816) and dominant (E X-linked dominant syndromes, p. 814) conditions. • Presumed polygenic conditions (e.g. neural tube defects, pervasive developmental disorders). • Mitochondrial disorders, maternally inherited [e.g. myoclonic epilepsy with ragged red fibres [MERRF]]. Central nervous system malformations of unknown aetiology About 60% of all CNS malformations do not have a known genetic or exogenous cause. The types of malformation seen indicate the timing of the causative event, but not its nature (see Table 17.2). Table 17.2 Types of malformation and timing of the causative event

Timing (in gestation)	CNS event	Malformation
3–7wks	Dorsal induction	Anencephaly, encephalocele, meningomyelocele, other neural tube closure defects
5–6wks	Ventral induction	Prosencephalies and other faciotelencephalic defects
2–4mths	Neuronal proliferation	Microcephaly or macrocephaly
3–5mths	Neuronal migration	Gyrus anomalies and heterotopias
6 mths (to first year of life)	Neuronal organization	Myelination. Disturbed connectivity (dendrite/synapse formation). Disturbed proliferation of oligodendrocytes and myelin sheets

Aetiology External prenatal factors (E Non-genetic causes of intellectual disability, p. 818.) Infection; exposure to medication, alcohol, drugs, and toxins; maternal illness (diabetes, hypothyroidism, hypertension, malnutrition) and gestational disorders. These factors are particularly damaging in the early stages of fetal development during blastogenesis or organogenesis. Perinatal factors Occurring around the time of delivery. Neonatal septicaemia; pneumonia; meningitis/encephalitis; other congenital infections; problems at delivery (asphyxia, intracranial haemorrhage, birth injury); other newborn complications (respiratory distress, hyperbilirubinaemia, hypoglycaemia). Postnatal factors Occurring in the first years of life. CNS infections, vascular accidents, tumours; causes of hypoxic brain injury (e.g. submersion); head injury (e.g. RTAs, child abuse); exposure to toxic agents; psychosocial environment (i.e. deprivation). Other disorders of unknown aetiology (E Disorders of unknown aetiology, p. 819.) These include: cerebral palsies, epilepsy, ASD, and childhood disintegrative disorders.

796 Chapter 17 Intellectual disability Establishing the cause This requires a comprehensive history from the parents, examination of antenatal and perinatal records, and physical examination of the child. Factors in the history • Family history—parents: ages; consanguinity; medical history; any previous pregnancies (including abortions, stillbirths). Wider family: any history of ID; specific cognitive impairments; congenital abnormalities; neurological or psychiatric disorders. • Gestational history—general maternal health and nutrition; maternal infections; exposure to medication, drug and alcohol use, toxins, radiation; chronic medical conditions; history of pre-eclampsia, abnormal intrauterine growth or fetal movements. • Birth of child—gestational age; whether multiple pregnancy (birth order); duration of labour; mode of delivery; any complications; any placental abnormalities. Examination of birth records (Apgar scores, weight, length, head circumference). • Neonatal history—need for special care (respiratory distress, infections, hypoglycaemia, hyperbilirubinaemia), baby checks (physical examination, Guthrie test). • Childhood history—weight gain, growth pattern, feeding pattern, sleeping pattern, early developmental milestones. History of childhood illnesses (especially CNS infections or seizures, metabolic/endocrine disorders) and accidents. General systemic enquiry. Physical examination • Look for evidence of any dysmorphic features, and note whether these are seen in close relatives (e.g. skin—pigmentation, dermatoglyphs; facial features; musculoskeletal abnormalities). • Full physical examination of all systems, including neurological examination for localizing signs. • If

suggested by the history/examination, ophthalmic and audiology examinations should be arranged.

Investigations • Standard tests will include FBC, U&Es, LFTs, TFTs, glucose, infection screening (blood and urine), and serology (ToRCH—toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus; HIV). • Where dysmorphic features are evident, or physical signs indicate, arrange X-rays of the skull, vertebrae, chest, abdomen, hands, feet, and long bones; cardiac/abdominal ultrasound. • If a metabolic disorder is suspected (e.g. progressive course), arrange screening tests of blood and urine. • If a genetic disorder is suspected, tests may include array comparative genomic hybridization (CGH), karyotyping, or more specific genetic testing (e.g. FraX DNA testing). • Other more detailed investigations may include neurophysiological tests (EEG, evoked potentials), neuroimaging (cranial ultrasound, CT/MRI, functional imaging), (neuro)pathological examination (fibroblast culture; biopsies—muscle, skin, rectum).

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798 Chapter 17 Intellectual disability The process of assessment When a person with ID presents to services because of a particular problem (e.g. 'challenging behaviour' or mood disturbance), the task for the clinician is to determine the underlying cause and to consider any predisposing, precipitating, and perpetuating factors. Causation will often be multifactorial, and because of this, a structured assessment approach is best. Some aspects of assessment may be well documented (e.g. the aetiology of the ID), particularly when the patient is an adult. Any diagnostic formulation should always take note of previous assessments and highlight what further assessments may be helpful. It is always useful to consider any protective factors which can potentially be harnessed or used to aid improvement. Assessment of the nature and severity of the intellectual disability • Intellectual impairment—assessed using standardized psychometric tests (e.g. Wechsler scales). There are often important differences in subscale scores (e.g. verbal vs performance IQ). • Severity of ID—using ICD-10 or DSM-5 criteria (E Classification, p. 790). • Disabilities—assessments of functioning [e.g. Vineland Adaptive Behaviour Scales, American Adaptive Behavior Scales, Hampshire Assessment for Living with Others (HALO)]. • Impact—assessment of quality of life and life experiences (e.g. Life Experiences Checklist). • Aetiology—E Establishing the cause, p. 796.

Assessment of the current problem • Full physical examination—this may identify undiagnosed problems, which the patient may be unable to communicate. • MSE—E Psychiatric comorbidity in intellectual disability, p. 826. Mental illness (which may go unrecognized and untreated) can be a causative or a complicating factor in many presentations. • Environmental and social factors—in addition to assessment of the patient, attention should also be focused on the patient's living situation, relationships, activities, and current stressors, noting particularly any recent changes.

Current support network Assessment will involve not only talking to the patient, but also gathering information from previous documentation (including previous diagnoses and current treatments) and talking to the family and to any carers and to any other support services or education services involved. The aim is to view the current problem in the light of past experiences, known problems, and current situational factors. A longitudinal approach is advised (i.e. does the current presentation reflect a recurrent problem, or is it part of progressive functional decline, or does it represent a new, unidentified problem or an unmet need?). It is useful to document the current supports received by the patient and any important contacts for future reference.

The process of assessment Needs assessment Should it be the case that the person's needs have changed, then there may be a statutory responsibility to undertake a formal 'needs assessment',

taking into account the wishes of the person (if they have capacity to make the kinds of decisions required) and others involved in care provision. This includes social care, educational, and healthcare needs. Increasingly, more joint work is being done between psychiatry of ID departments and social work departments. As a result, it is likely that you will be more involved in discussions and assessments of patients, with a view to deciding on what their overall care needs are, in addition to their particular psychiatric needs. Naturally, this will be in an MDT but will include consideration of:

- Level of supervision by nursing/care staff, e.g. day and night staffing ratios, sleeping vs waking night cover, same building or next door building.
- Layout of the building.
- Location of the building.
- Compatibility of different patients if being considered for group accommodation.

800 Chapter 17 Intellectual disability Communicating with people with intellectual disabilities Up to 90% of people with ID have some form of difficulty with communication, the severity of which can vary. Coupled with this, there is a much higher level of sensory impairment among people with ID than in the general population. These difficulties can make effective communication with someone with ID more complex and require a more skilled approach. It is important to introduce yourself fully to the person you are assessing and in a way they understand.

Borderline/mild intellectual disability People with borderline, or even mild, ID can appear to understand conversations and have developed sufficient social skills to allow them to mask their deficit in communication skills. They may be too embarrassed to reveal their lack of understanding, therefore meaning they leave the consultation without understanding what has been said to them. It is important to use simple, everyday language in short sentences and to check the person understands. As with standard assessments, it is important to start the consultation with open questions, but the move to more closed questions may be quicker as the person may find it easier to answer questions where there is less doubt about them. The interview may take longer than you expect, but try not to hurry things along by trying to finish sentences or try and guess what the person is trying to say. It helps to be definite around the length of the consultation if you need to be—this avoids the person feeling they have done something wrong when the consultation ends, particularly as they may not pick up on subtle signs that the consultation is ending.

Moderate/severe intellectual disability As the severity of the ID worsens, it is likely that communication skills will be more limited, although it is important not to assume this. Non-verbal signs, such as gestures and facial expressions, are even more important with people with a moderate/severe ID. For example, using a thumbs up/ thumbs down to judge good or bad and using a serious facial expression when talking about something serious can be helpful.

Environmental considerations Bearing in mind the level of sensory impairment, carrying out an assessment in a quiet environment with limited distractions will be of benefit to all parties involved. This ensures, even with sensory impairment, the best chance of the person hearing or understanding you. The lack of distractions also ensures you are able to hold the person's attention for as long as possible.

Collateral history Most people with ID will attend consultation with a support worker or carer, either formal or informal. It is important to ask consent from the person to have their carer present during the consultation. It can be very helpful to have a carer present, as they will likely be able to assist with

Communicating with people with intellectual disabilities communication, as well as provide a collateral history. One should, however, take care to keep the patient with ID the focus of the consultation, avoiding the temptation to speak only to the carer. You should also give the patient

the opportunity to speak to you alone if they wish, as they may have concerns they do not wish to disclose in front of their carer. They will also be able to discuss further with the person once they leave the room and remind them of the information they have been given. Asking about symptoms It can be difficult at times to distinguish what symptoms are present in someone with ID due to communication difficulties. It may be helpful to identify any changes in behaviours or routines, and to look for things which are different from the norm for that person. It is important not to allow yourself to assume that current symptoms can be attributed to a develop mental disorder or other diagnosis (diagnostic over-shadowing). In some circumstances where a person has more severe communication difficulties, the diagnosis of mental illness may be based more on observations and collateral histories. Communication systems Some people with ID have specific systems they use to communicate, nor mally in the form of communication aids. An example of this is the 'Picture Exchange Communication System' (PECS). This is a system initially devel oped for children with autism, which is now used for people with ID. This system allows them to communicate through symbols in order to make themselves understood. Widgit is another way of communicating through symbols and can be used for people who are unable to understand written material (e.g. in leaflets). Widgit can be used to communicate with people who also struggle to understand more complex situations.

802 Chapter 17 Intellectual disability Considering management choices The therapeutic environment Provision of care and support should always be within an appropriate setting. Support may be: general (care provided by usual carers, schools, and community teams) and/or specific (addressing particular needs, e.g. special education, parental support groups, physical or psychiatric problems, mal adaptive behaviours). Although, in general, every effort will be made to sustain a 'normal' environment (remaining at home, integration into main stream schools, use of local community resources), often more specialized environments are necessary. Overcoming communication difficulties • Use of aids to overcome/improve sensory deficits (e.g. hearing aids, glasses). • Strategies for improving communication—PECS, symbol dictionaries, Makaton, sign language. • Because of often unique communication styles, it is important that family/carers who know the patient are available to assist/improve communication and that their expertise is shared among new staff. Factors influencing management choices • The nature of the problem (e.g. biological, psychological, social). • The degree and aetiology of the ID. • Comorbid physical conditions (which may restrict medication choices). • Situational factors (e.g. practicalities of instituting various treatment options, supports, ability to monitor progress). Admission to specialist environments Sometimes disabilities or problems may be too severe or too complex to be managed with standard community resources because: • The degree of ID or the specific cognitive impairments requires a well- structured, predictable environment that cannot be provided elsewhere. • The degree of physical impairment requires more intensive specialist nursing or a safer environment where medical care is close at hand (e.g. severe treatment-resistant epilepsy). • The severity of behavioural problems prohibits management at home (e.g. abnormally aggressive or disinhibited behaviour which constitutes a serious risk of harm to the patient or to others). • The person requires treatment for a comorbid psychiatric disorder, which has failed to respond to initial treatment or requires a review of medications. In the past, there was a preference for people with ID to remain as in patients for a prolonged period, but recently there has been a drive to try and reduce this (see Box 17.2).

Considering management choices Other reasons for admission may include • Respite placements to allow individuals and their families some relief from the intensity of long-term care. • Assessment of complex problems—to disentangle environmental from illness factors or where treatment requires close monitoring. • ‘Crisis’ admissions due to an acute breakdown of usual supports. Cautionary notes • Attributing treatment success to a particular intervention may miss the real reason for improvement, e.g. return of a familiar carer, a more structured environment (if admitted to a specialist centre), or treatment effects on an undiagnosed primary condition (e.g. an anticonvulsant used for aggressive behaviour may actually be treating underlying epilepsy). • Many conditions may run relapsing-remitting courses, leading to mistaken conclusions about the effectiveness of an intervention, which only becomes clear when symptoms return despite treatment. • Improvement (or worsening) of symptoms may reflect normal maturational processes or, conversely, further pathological degeneration. • Because of the wide variation in aetiology (genetic, environmental, psychological, social) and the complexity (and variable degrees) of cognitive impairment, most trials of treatment are, by nature, empirical. Most management plans will inevitably be individually tailored, and the current evidence base for many treatment modalities is limited. • People within inpatient units should undergo regular review of their care and treatment, particularly to assess if inpatient care is still required. Box 17.2 Winterbourne View Hospital In 2011, a television programme uncovered abuse of patients with ID by staff in an assessment and treatment unit in England, which prompted a Department of Health review¹ across NHS England, looking at the care and treatment of patients with ID. The review concluded that these patients were receiving poor care in inpatient facilities for too long a period and too far from home. As a result of this review, an initiative was launched to reduce the number of inpatients. Care and treatment reviews were also introduced, and NICE guidelines about challenging behaviour (Oct 2015; M <https://www.nice.org.uk/guidance/QS101>) and care and treatment (Jan 2017; M <https://www.nice.org.uk/guidance/QS142>) of patients with IDs were published [both accessed 11 July 2018]. 1 Department of Health: Transforming Care: A National Response to Winterbourne View Hospital. Dec 2012. M <http://www.rcpsych.ac.uk/pdf/final-report.pdf> [accessed 11 July 2018].

804 Chapter 17 Intellectual disability Treatment methods Behavioural treatments May be used to help teach basic skills (e.g. feeding, dressing, toileting) and establish normal behaviour patterns (e.g. sleep) or more complex skills (e.g. social skills, relaxation techniques, assertiveness training). Behavioural techniques may also be used to alter maladaptive patterns of behaviour (e.g. inappropriate sexual behaviour, pica, phobias). Pharmacological treatments (See Box 17.3.) Cautions • Comorbid physical disorders (e.g. epilepsy, constipation, cerebral palsy) increase the need to closely monitor adverse effects. • Atypical responses, such as i (or reduced) sensitivity and ‘paradoxical’ reactions, are more common; hence, low doses and gradual increases in medication are advisable. • The evidence base for many drug treatments is lacking, and many claims for efficacy are, at best, based on small, open, uncontrolled trials. Antipsychotics For the treatment of comorbid psychiatric disorders (e.g. schizophrenia and related psychosis) and acute behavioural disturbance. May also be effective in managing ASD, self-injury, social withdrawal, ADHD, and tic disorders. Should only be used under specialist guidance. Antidepressants Effective for the treatment of depression, OCD, and other anxiety disorders. They have also been used in the management of violence, self-injury, ‘non-specific’ distress, and other compulsive behaviours. Box 17.3 Standards for psychotropic drug prescribing The Royal College of Psychiatrists issued standards for psychotropic drug prescribing in patients with ID, partly in response to Winterbourne

View Hospital (see Box 17.2), due to concerns about patients receiving too much medication:1 • Indication and rationale for prescribing the psychotropic drug must be stated. • Consent to treatment procedures should be followed. • There should be regular monitoring of treatment response and side effects. • Review and evaluation of the need for continuation or discontinuation of the psychotropic drug should be undertaken on a regular basis (preferably every 3mths at a minimum). 1 Faculty of Intellectual Disabilities: Psychotropic Drug Prescribing for People with Intellectual Disability, Mental Health problems and/or Behaviour Which Challenges. Practice Guidelines RCPsych April 2016. M http://www.rcpsych.ac.uk/pdf/FR_ID_09_for_website.pdf [accessed 12 July 2018].

Treatment methods Anticonvulsants There is some evidence for the use of anticonvulsants in the treatment of episodic dyscontrol (e.g. carbamazepine), but their effectiveness may be due to better control of underlying epilepsy. Lithium Aside from the treatment of bipolar affective disorder and augmentation of antidepressant therapy, lithium may have some utility in reducing aggressive outbursts. Beta-blockers May be useful in conditions of heightened autonomic arousal (e.g. anxiety disorders), which may be at the root of aggressive behavioural disturbance. Stimulants For the treatment of ADHD (e.g. methylphenidate) (E Attention-deficit/ hyperactivity disorder 2: medication, p. 670). Opiate antagonists May be effective in the treatment of repetitive self-injury (e.g. naltrexone). Anti-libidinal drugs Used in the treatment of sexual offending (e.g. cyproterone acetate and medroxyprogesterone, which reduce testosterone levels) (E Sexual of fences 2, p. 740), under specialist guidance. Cognitive therapies and cognitive behavioural therapy For borderline, mild, or moderate ID, cognitive approaches may be adapted to the level of intellectual impairment and the patient's style of communication. These may be effective in the teaching of problem-solving skills, the management of anxiety disorders and depression, dealing with issues of self-esteem, anger management, and treatment of offending behaviours (e.g. sex offenders). Psychodynamic therapies May be helpful in addressing issues of emotional development, relationships, and adjustment to life events (e.g. losses, disabilities, and bereavement). The range of approaches varies from basic supportive psychotherapy to more complex group and family therapies.

806 Chapter 17 Intellectual disability Down's syndrome Down's syndrome (trisomy of chromosome 21) is the most common genetic cause of ID (1:800–1:1000). It is characterized by intellectual impairment and associated characteristic facies and habitus. Although Down's syndrome is diagnosed at birth, ID only becomes evident at the end of the first year of life, with subsequent delayed developmental milestones. The IQ in adults is most often below 50 (range: low to high/moderate ID). Those who survive into their 40s and 50s show pathological brain changes similar to Alzheimer's disease. Aetiology Risk factors for giving birth to a child with Down's syndrome are: maternal age over 40yrs; a previous child with the syndrome; and Down's syndrome in the mother (although pregnancy is rare). Incidence per 1000 living births is 70.5 for a woman under 25, 0.7 under the age of 30, 5.0 under 35, 25 under 40, and 34.6 over the age of 45. Most children with Down's syndrome (70–80%) are born to mothers under the age of 35 (due to higher number of pregnancies in younger women). Genetics Full trisomy 21 (non-disjunction) in 95% of cases. Robertsonian translocations in 5% (of which 45% show fusion—usually 14 and 21; also 13/15/ 22 and 21 described). Mosaicism (a mixture of normal and trisomic cell lines) in 2–5%—IQ can be in the 70s, and physical abnormalities may be less marked. Clinical features • General—short stature (mean 1.4–1.5m), overweight (30%), muscular hypotonia. • Head and

neck—brachycephaly and reduced anteroposterior (AP) diameter, maxilla reduced more than the mandible, underdeveloped bridge of the nose, eyes close together, Brushfield's spots (grey or very light yellow spots of the iris), epicanthic fold, low-set ears, high-arched palate, protruding tongue, instability of atlanto-axial joint, narrowed hypopharynx (may lead to sleep apnoea). • Congenital heart defects—(50%), e.g. atrial or ventricular septal defect, mitral valve disease, patent ductus arteriosus. • Congenital GI abnormalities—oesophageal atresia, Hirschsprung's disease, umbilical and inguinal hernia. • Hands—short, broad hands with a single palmar crease (simian crease), syndactyly (webbed fingers), clinodactyly (incurving of fingers), and altered dermatoglyphics. • Eye defects—strabismus (20%), myopia (30%), blocked tear ducts, nystagmus, late-life cataracts, keratoconus. • Hearing defects—structural anomalies may lead to recurrent otitis media, sensorineural deafness. • Immunological abnormalities—raised immunoglobulin G (IgG) and immunoglobulin M (IgM), lowered T-lymphocytes.

Down's syndrome • Endocrine abnormalities—thyroid dysfunction (hypothyroidism—20%), diabetes. • CNS abnormalities—reduced brain weight (10–20%), reduced gyri, cortical thinning, underdeveloped middle lobe of the cerebellum, reduced neuronal numbers in the cerebellum/locus caeruleus/basal forebrain, reduced cholinergic neurons, neuropathological changes similar to Alzheimer's disease (in those over 40yrs), epilepsy (5–10%). • Abnormal sexual development—♂: normal course; delayed puberty; problems with spermatogenesis (unless mosaic). ♀: normal onset of menstruation; fertile, but problems with ovulation and follicular growth; early menopause. • Psychiatric comorbidity—in 18% of children and 30% of adults with Down's syndrome (usually depression 10%; less commonly bipolar disorder, OCD, Tourette's, schizophrenia, i risk of autism). Dementia in Down's syndrome While dementia of the Alzheimer's type (DAT) is the most common type in Down's syndrome, all types of dementia can occur. There is an i risk of DAT due to genetic factors (the amyloid precursor protein gene on chromosome 21 is implicated in early-onset Alzheimer's disease). Unfortunately, the diagnosis is often difficult, given the premorbid cognitive deficits and communication difficulties. The crucial element in diagnosis is establishing a history of change from an informant who has known the patient over a sufficient period as to be able to make a useful comparison. Assessment • Full history, focusing on previous abilities, presentation, and behaviour. • Exclusion of other physical/psychiatric explanations, e.g. sensory loss, delirium, hypothyroidism, depression. • Use of a standardized cognitive assessment battery, either to act as a baseline for decline or response to treatment. • Full blood investigations, including FBC, U&Es, ESR, LFTs, TFTs, glucose, folate and vitamin B12, and serum drug levels if relevant. • Consideration of CT/MRI brain or EEG, if indicated. Management • Treat all reversible additional factors. • Optimize communication: use of pictures, communication dictionary, etc. (E Considering management choices, p. 802). • Liaison with psychology colleagues for potential behavioural management. • Consideration of anticholinesterase inhibitors, but titrating at a significantly slower rate than normal (e.g. donepezil 5mg nocte for 4–6wks before increasing the dose). Seek advice from local experts. • Appropriate placement, considering client mix, age group, and range of available activities.

808 Chapter 17 Intellectual disability Deletions and duplication syndromes Angelman ('happy puppet') syndrome Microdeletion (60–75% of cases); karyotype 15q11–q13; incidence 1:10,000; a contiguous gene syndrome (the complement of PWS), with 80% due to deletion of maternally derived chromosome 15, 2% paternal uniparental disomy (pUPD), and the remainder due to direct mutations. Clinical features—ataxia (jerky limb movements, gait problems); epilepsy (86%);

paroxysms of laughter; absence of speech; facial features (blond hair, blue eyes, microcephaly, flat tened occiput, long face, prominent jaw, wide mouth, widely spaced teeth, thin upper lip, mid-facial hypoplasia); severe/profound ID; other behaviours (hand flapping, tongue thrusting, mouth movements); other problems [upper respiratory tract infections (URTIs), ear infections, obesity]. Beta-thalassaemia Mental retardation. Small deletion; karyotype 16pter- p13.3 (cryptic terminal deletion). Clinical features—ID. Cri-du-chat Partial monosomy; karyotype 5p- (varies from deletion of a small band at 5p15.2 to the entire arm of 5p); usually sporadic, occasionally inherited; incidence 1:35,000. Clinical features—‘cat-like’ cry (possibly due to abnormal laryngeal development), microcephaly, rounded face, hypertelorism, micrognathia, dental malocclusion, epicanthic folds, low-set ears, hypotonia, severe/profound ID. Puberty occurs normally, and some may survive to adulthood. di George (velo-cardio-facial) syndrome Microdeletion; karyotype 22q11.2; incidence 1:2000. Clinical features—50% have ID (mild: two-thirds; moderate: one-third), cardiac abnormalities [75%: Fallot tetralogy, ventricular septal defect (VSD), interrupted aortic arch, pulmonary atresia, truncus arteriosus], facial features (microcephaly, cleft palate/submucous cleft, small mouth, long face, prominent tubular nose, hypoplasia of adenoids—nasal speech, bulbous nasal tip, narrow palpebral fissure, minor ear abnormalities, small optic discs/tortuous retinal vessel/cataracts), hypocalcaemia (60%—seizures, short stature, hearing problems, renal problems, inguinal/umbilical hernia), hypospadias (10% of ♂), long and thin hands (hypotonia and hyperextensible fingers), associated behavioural and psychiatric disorders (including schizophrenia, blunted/inappropriate affect). Prader-Willi syndrome (PWS) Microdeletion; karyotype 15q11-q13; incidence 1:40,000; the complement of Angelman syndrome; 75% due to deletion of paternally derived chromosome 15, 25% due to maternal uniparental disomy (mUPD) (i.e. inheritance of two genes from the same parent), ♂:♀ = 4:3. Essence—the striking feature of PWS is massive hyperphagia with associated compulsive food-seeking, and consequent marked obesity. The hyperphagia may be such that questions of how to appropriately limit the person’s access to food must be addressed, sometimes requiring consideration of measures under capacity/incapacity legislative frameworks. Clinical features—neonates: hypotonia, sleepiness, unresponsiveness, narrow bifrontal diameter, triangular mouth (feeding difficulties and swallowing problems), strabismus, acromicria (shortness of extremities). Childhood/adolescence: short stature, hypogonadism (cryptorchidism, micropenis; amenorrhoea), behavioural disorders (overeating and obesity, self-injurious behaviour), mild to moderate ID, speech abnormalities, sleep

Deletions and duplication syndromes disorders. Affective psychoses are associated, particularly with the mUPD genotype. Associated features—small hands and feet, cleft palate, almond-shaped eyes, strabismus, incurved feet, clubfoot, congenital hip dislocation, abnormalities of the knees and ankles, scoliosis. Other physical problems—diabetes, GI problems (obstruction, duodenal ulcer, rectal prolapse, gall stones), heart disease, respiratory problems (asthma, cor pulmonale), renal calculi, hearing deficits, hypothermia. Rubenstein-Taybi syndrome Microdeletion of the gene encoding human cAMP-regulated enhancer binding protein; karyotype 16p13.3; incidence 1:125,000. Clinical features—ID and dysgenesis of the corpus callosum. Broad thumbs and great toes; persistence of fetal finger pads; facial features (short upper lip, pouting lower lip, maxillary hypoplasia, beaked nose, slanted palpebral fissure, long eyelashes, ptosis, epicanthic fold, strabismus, glaucoma, iris coloboma); cardiac problems (pulmonary stenosis and hypertension, mitral valve regurgitation, patent ductus arteriosus); propensity to keloid formation; genitourinary features (hypoplastic kidneys, cryptorchidism, small scrotum); GI problems (constipation, megacolon); collapsible larynx (leading to sleep apnoea); epilepsy (25%); behavioural problems

(sleep problems, stereotypies, e.g. rocking, self-injurious behaviour). Smith–Magenis syndrome Deletion in 17p11.2; incidence 1:50,000. Clinical features—moderate ID; facial features (brachycephaly, broad face, flattened mid-face, strabismus); myopia; short, broad hands; upper limb deformity; insensitivity to pain. Behavioural problems—‘self-hugging’ posturing, aggression, self-injury, hyperactivity, severe sleep problems, other autistic features. Williams syndrome Small deletion; karyotype 7q11.23 (possibly gene for elastin or protein kinase—LIMK1); 1:15,000 live births; may also be related to excessive maternal vitamin D intake. Clinical features—hypercalcaemia (in 750%) with supraaortic stenosis and unusual facies. Neonates: may be irritable, have feeding problems, and failure to thrive. Childhood: growth retardation, ‘elfin’ facial features, hoarse voice, premature wrinkling and sagging of the skin, cardiovascular anomalies (e.g. supraaortic stenosis), urinary tract abnormalities (asymmetrical kidneys, nephrocalcinosis, bladder diverticuli, urethral stenosis), pulmonary artery stenosis, mild to moderate ID (verbal often better than visuospatial and motor abilities). Often there is abnormal attachment behaviour (manifest as anxiety, poor peer relationships, hypersensitivity, or conversely as social disinhibition, excessive friendliness). Wolf–Hirschhorn syndrome Partial monosomy; karyotype 4p–. Clinical features—severe ID; many survive to adulthood.

810 Chapter 17 Intellectual disability Autosomal dominant syndromes Noonan’s syndrome Occurring in 1:1000–1:2000; ♂ = ♀. Initial description of nine children seen in the Congenital Heart Disease Clinic who shared characteristic facies, anterior chest wall deformities (pectus carinatum or excavatum), and short stature. While a number of genes (including PTPN11, SOS1, and KRAS) have been identified, which can cause Noonan’s syndrome, it remains a clinical diagnosis. Clinical features—varying degree of ID (from none to severe), short stature (80%), cardiac abnormalities (>80%), hepatosplenomegaly (25%), distinctive facies. The following group of disorders are also termed phakomatoses—a variety of conditions of ectodermal origin with neurocutaneous signs. Although von Hippel–Lindau syndrome is not associated with ID, it is included for completeness. Tuberous sclerosis (TSC) Occurring in 1:7000–10,000; ♂ = ♀. Clinical features—varying degree (usually severe) of ID (50%), seizures (e.g. ‘Salaam attacks’ and other types, in 90%), hamartomas of the CNS (including the retina), as well as ependymomas and astrocytomas, facial angiofibroma, adenoma sebaceum, depigmented skin patches (‘ash leaf spots’ in 96%), shagreen patches, depigmented naevi, subcutaneous nodules, ‘café-au-lait’ spots, fibromas of the nails, pitted tooth enamel, hypoplasia, and occasionally tumours of the heart (rhabdomyeloma, hamartoma), kidney problems (Wilms’ tumour, renal cysts), olfactory hamartomas, hypertension, and aortic aneurysm. Subtypes—TSC1: 1:12,000; associated with a gene (for hamartin—believed to be tumour-suppressing) near the ABO blood group locus on chromosome 9 (9q34—40% of cases); TSC2: associated with a gene for tuberlin (a guanosine triphosphatase-activating protein, also believed to be tumour-suppressing) on chromosome 16 (16p13.3–), more psychiatric and behavioural problems; TSC3: a rare translocation of a gene on chromosome 12. Neurofibromatosis Type 1 (NF1, von Recklinghausen’s disease)—occurring in 1:3000; ♂ = ♀. Autosomal dominant condition, responsible gene on chromosome 17 (75% spontaneous mutations). Clinical features—café-au-lait spots, freckling, dermal neurofibromas, nodular neurofibromas, Lisch nodules; associated with mild intellectual disability in 75%. Type 2 (NF2)—occurring in 1:35,000. Autosomal dominant condition, responsible gene on chromosome 22. Clinical features—bilateral vestibular schwannomas, café-au-lait spots, juvenile posterior subcapsular lenticular opacities. Sturge–Weber syndrome Caused by spontaneous genetic mutation in unknown location. Clinical features—‘port-

wine stain', typically covering part of the forehead and at least one eyelid, angiomas of the meninges in the temporal and occipital areas on the same side as the port-wine stain. Associated to varying degrees with ID. Epilepsy is the most common early problem, often starting before the age of 1. Hemiparesis may develop, usually contralateral to the port-wine stain. Buphthalmos (bulging of the eye) and glaucoma are common in the affected eye.

Autosomal dominant syndromes
von Hippel-Lindau (VHL) syndrome A rare genetic condition caused by a mutation of the VHL tumour suppressor gene on chromosome 3p; 80% inherited, 20% new mutation. Symptoms caused by angiomas in various areas of the body. Clinical features—renal cysts/carcinomas, pheochromocytomas, CNS haemangioblastomas, pancreatic cysts/tumours (can be neuroendocrine), subretinal haemorrhages secondary to retinal vessel tortuosities/aneurysms. Not associated with ID.

812 Chapter 17 Intellectual disability **Autosomal recessive syndromes** These conditions include some of the lysosomal storage diseases, e.g. mucopolysaccharide storage—Hurler syndrome, Sanfilippo disease, sphingolipid storage—Tay-Sachs disease, Niemann-Pick disease (sphingomyelins), glycoprotein storage—sialidosis; phenylketonuria; and rare disorders such as Laurence-Moon syndrome and Joubert syndrome.
Phenylketonuria A preventable cause of severe ID, due to deficiency of phenylalanine hydroxylase (long arm of chromosome 12), leading to phenylalaninaemia and phenylketonuria; incidence 1:10,000; diagnosed postnatally ('Guthrie test'). Clinical features—fair hair/skin and blue eyes (lack of pigment—tyrosine deficiency), neurological signs (stooped posture, broad-based gait, low tone and reflexes, tremor, stereotyped movements). Behavioural problems—hyperactivity, temper tantrums, perseveration, echolalia. Management—supervised early dietary restriction of phenylalanine. Prognosis—even with dietary treatment, lower-than-average IQ.
Sanfilippo disease Due to disorders of the breakdown of heparan sulfate, of which there are four subtypes (types A-D). Incidence 1:200,000. Clinical features—severe ID, claw hand, dwarfism, hypertrichosis, hearing loss, hepatosplenomegaly, biconvex lumbar vertebrae, joint stiffness. Behavioural problems—restlessness, sleep problems, challenging behaviour. Aetiology—type A (most severe, most common) mapped to 17q25.3 (heparan sulfate sulfatase); type B 17q21 (N-acetyl- α -D-glucosaminidase); type C on chromosome 14 or 21 (acetyl-CoA- α -glucosaminide-N-acetyltransferase); type D 12q14 (N-acetyl- α -D-glucosamine-6-sulfatase). Prognosis—poor, many die between 10 and 20yrs of respiratory tract infections.
Hurler syndrome Due to deficiency in α -L-iduronidase (4p16.3); incidence 1:100,000. Clinical features—progressive ID (eventually severe/profound), skeletal abnormalities (short stature, kyphosis, flexion deformities, claw hand, long head, characteristic facial appearance), hearing loss, respiratory and cardiac problems, hepatosplenomegaly, umbilical/inguinal hernia. Prognosis—poor, some survive to 20s; may benefit from allogeneic bone transplantation.
Laurence-Moon syndrome Associated with multiple loci (11q13, 11q21, 15q22, 3p13); prevalence 1:125,000–160,000 (higher in Bedouins of Kuwait and Newfoundland). Also known as Laurence-Moon-Biedl syndrome (incorporating Bardet-Biedl syndrome which shares clinical features, but additionally there is central obesity and polydactyly). Clinical features—mild to moderate ID, short stature, spastic paraparesis, hypogenitalism (most ♂ are infertile), night blindness (due to red cone dystrophy), non-insulin-dependent diabetes mellitus (NIDDM), renal problems (diabetes insipidus, renal failure).
Joubert syndrome Exceptionally rare, no loci identified, but recessively inherited. Clinical features—severe ID, characteristic hyperpnoea ('panting like a dog'), cerebellar dysgenesis, hypotonia, ataxia, tongue protrusion, facial spasm, abnormal eye

movements, cystic kidneys, syndactyly/poly dactyly. Behavioural problems—self-injury. Prognosis—poor, no specific treatments.

Autosomal recessive syndromes **Gaucher's disease** Most common of the lysosomal storage diseases. Caused by deficiency of glucocerebrosidase, leading to accumulation of glucosylceramide, most commonly in the spleen, liver, lung, bone, and brain. Type I—the brain is unaffected, onset later in adulthood; types II and III—associated with ID, with type II being the most severe. Prognosis—type I, close to normal; type II, children usually die by age of 2yrs; type III, adolescence to adulthood. Treatment—enzyme replacement and bone marrow replacement both used in the treatment of types I and III. Unfortunately, there is no treatment for the neurological effects in types II and III.

814 **Chapter 17 Intellectual disability** **X-linked dominant syndromes** **Fragile X syndrome** A common inherited cause of ID, affecting 1:4000 ♂ and 1:8000 ♀, with X-linked dominant transmission. Penetrance is low, but greater in ♂ than ♀ (due to 'protective' effects of the second normal X chromosome in ♀). Gene sequence has been cloned³ and designated FMR-1. The syndrome is associated with a large sequence of triplet repeats (CGG)_n at a fragile site on the X chromosome (Xq27.3). In affected ♂, $n > 230-1000+$; in transmitting ♂ and obligate ♀, $n = 43-200$; and in the general population, $n = 6-54$ (mean 30). Clinical features—variable, subtle, and often cannot be detected before adulthood. May include: large testicles and ears, smooth skin, hyperextensible fingers, flat feet, mitral valve prolapse, inguinal and hiatus hernia, facial features (long, narrow face with underdevelopment of the mid-face, macrocephaly), epilepsy (725%), variable ID (borderline to profound). Behavioural features—appear to be similar to those seen in ADHD and autism: hand flapping/waving, repetitive mannerisms, shyness, gaze avoidance, poor peer relationships, communication difficulties (e.g. delayed language development, conversational rigidity, perseveration, echolalia, palilalia, cluttering, and over-detailed/circumstantial speech), psychiatric problems (e.g. prominent depression/anxiety). Many of the features of fragile X also overlap with those of autism, although debate is ongoing as to the exact nature of the relationship. Note: general domestic and daily living skills may be excellent. Brain imaging—reduced posterior cerebellar vermis, enlarged hippocampus and caudate nuclei, enlarged ventricles. Other disorders with 'fragile' sites Two other fragile sites have been found on the X chromosome. The original 'fragile X' site has hence been designated 'FRAX A'. FRAX E, caused by FMR-2 mutation, is also associated with mild ID, with an incidence of 1:100,000 and 200-1000 triplet repeats. FRAX F has not (yet) been associated with any disorder. Another fragile site has been located on chromosome 16 (FRA 16), associated with a large GCC triplet expansion—but no specific clinical disorder. **Rett's syndrome** A pervasive developmental disorder (E Pervasive developmental disorders, p. 820) almost exclusively affecting ♀, with an incidence of 1:10,000. Initially described by the Austrian physician Andreas Rett in 1966,⁴ but only fully recognized after a second paper in 1983.⁵ Clinical features—initially normal development, followed by four stages: 3 Verkerk AJ, Pieretti M, Sutcliffe JS, et al. (1991) Identification of a gene (FMR-1) containing a CGG repeat coincident with a breakpoint cluster region exhibiting length variation in fragile X syndrome. *Cell* 65:905-14. 4 Rett A (1966) Ueber ein eigenartiges hirnatrophisches Syndrom bei Hyperammoniamie im Kindesalter. *Wien Med Wschr* 116:723-8. 5 Hagberg B, Aicardi J, Dias K, et al. (1983) A progressive syndrome of autism, dementia, ataxia, and loss of purposeful hand use in girls: Rett's syndrome: report of 35 cases. *Ann Neurol* 14:471-9.

X-linked dominant syndromes

1. Early onset/developmental arrest.
 2. Rapid destructive/regressive.
 3. Plateau (or pseudo-stationary) and (4) late motor deterioration:
 - Stage 1 Onset usually 6–18mths. May be delays in gross motor skills. Infants may show decreased eye contact and interest in toys. The typically described hand-wringing and decreased rate of head growth may also be apparent.
 - Stage 2 Onset usually between 1 and 4yrs. Purposeful hand movements and spoken language are lost. Stereotypical hand movements, including wringing, washing, clapping, or tapping, are often seen. Emergence of some autistic symptoms and a worsening gait may be seen.
 - Stage 3 Onset usually before age 10, and can last for most of life. Seizures and motor problems more prominent. Some improvement in behaviour, with more interest in others and surroundings, and some improvement in communication skills.
 - Stage 4 This stage can last for decades and is typified by gradual worsening in mobility, with scoliosis, spasticity, and muscle weakness.
- Aetiology** Mutations in the MECP2 gene on the X chromosome are present in the majority of girls with Rett's syndrome. Mutations on the CDKL5 gene have also been implicated in a variant of Rett's syndrome with notably early-onset of seizures.
- Prognosis**—poor, with continued motor deterioration and usually severe intellectual disability.
- Aicardi syndrome** Rare (only 200 reported cases—all ♀); dysgenesis of the corpus callosum and cerebrum, with severe ID; prognosis poor (often death in infancy). Clinical features—microcephaly, facial asymmetry, low-set ears, eye lesions (chorioretinal lacunae), hypotonia, scoliosis, epilepsy. Behavioural problems—25%: aggression, lack of communication, tiredness/sleep problems, self-injurious behaviour.

816 Chapter 17 Intellectual disability X-linked recessive syndromes These include other lysosomal storage diseases, e.g. mucopolysaccharide storage—Hunter syndrome; trihexosylceramide storage—Fabry disease, and other extremely rare conditions such as Lesch-Nyhan syndrome and oculocerebrorenal syndrome of Lowe.

Hunter syndrome Caused by iduronate sulfatase deficiency (mapped to Xq27–28); incidence 1:100,000 (more common in ♂ Ashkenazi Jews: 1:34,000). Symptoms are caused by build-up of glycosaminoglycans (GAGs) in a variety of body tissues. Only 20% have complete depletion of iduronate sulfatase, and two subtypes are recognized: type A—progressive ID and physical disability, with death before age 15yrs; type B—milder form, with minimal intellectual impairment and better prognosis.

Clinical features—short stature, distinctive coarse facies 'gargoylism', prominent forehead, enlarged tongue, flattened bridge of the nose, enlarged head, degenerative hip disease, joint stiffness, claw hand, chest deformities (pes cavus or excavatum), cervical cord compression, hepatosplenomegaly, hearing loss, breathing obstruction, developmental delay, eye defects (retinitis pigmentosa, papilloedema, hypertrichosis), umbilical/inguinal hernia.

Lesch-Nyhan syndrome An extremely rare X-linked recessive condition, due to a mutation in the HPRT gene (hypoxanthine phosphoribosyl transferase) on the short arm of chromosome Xq26–27, with a nearly total loss of the enzyme, leading to hyperuricaemia. Incidence 1:380,000. Prognosis is poor, and most affected individuals die in early adulthood.

Clinical features—children appear healthy at birth; dystonias become apparent around 3–4mths with delayed developmental milestones; later there is development of spasticity, choreoform movements, and transient hemiparesis (which may be misdiagnosed as cerebral palsy); variable degree of ID (usually severe); microcephaly is common; 750% develop epilepsy. Behavioural

problems—around age 2yrs (sometimes not until adolescence), self-mutilating behaviours may be seen (biting of lips, inside of mouth, fingers). Sometimes there is an episodic pattern, and some may show a reduction in frequency and severity after the age of 10yrs. May be associated with verbal and physical aggression. There is no clear cause for this behaviour—CNS findings include a reduction in DA in the basal ganglia and at synaptic terminals (but not in the cell bodies of the substantia nigra), with other monoaminergic systems apparently intact. Management—even treating hyperuricaemia does not appear to reduce behavioural problems; however, there is some evidence for the use of SSRIs. Oculocerebrorenal syndrome of Lowe Very rare X-linked recessive condition (Xq24–26); incidence 1:200,000. Clinical features—moderate to severe ID (up to 25% have normal IQ), short stature, hypotonia, epilepsy (730%), eye problems (e.g. congenital cataracts), renal problems (tubular dysfunction). Behavioural problems—temper tantrums, hand-waving movements, self-injury (770%—especially in early adolescence).

Sex chromosome disorders Sex chromosome disorders Turner's syndrome Sex chromosome monosomy; karyotype 45,XO; ♀ phenotype; 1:10,000 live births; generally normal IQ, with ID rare, although there may be specific deficits of visuospatial learning. Trisomy X Sex chromosome trisomy; karyotype 47,XXX; 1:1000 ♀ births. Clinical features—slight increase in height, 770% have intellectual disorder (usually mild), some evidence of reduced fertility (children have normal karyotypes), possibly incidence of schizophrenia. Klinefelter's syndrome Sex chromosome trisomy; karyotype 47,XXY; 1:1000 ♂ births (50% due to paternal and 50% maternal non-dysjunction). Clinical features—variable degree of development of secondary sexual characteristics, with hypogonadism, scant facial hair (90%), and gynaecomastia (50%). Taller than average (74cm), asthenic body build, median IQ 790 with skewed distribution—most in the 60–70 range, uncertain association with psychiatric disorders. XYY ♂ Sex chromosome trisomy; karyotype 47,XYY; 1:1000 ♂ births. Clinical features—controversial suggestion of higher incidence in prison populations, IQ may be slightly lower than average, behavioural problems commonly seen.

818 Chapter 17 Intellectual disability Non-genetic causes of intellectual disability Fetal alcohol spectrum disorder (FASD) One of the major causes of ID; incidence 0.2–3 per 1000 live births. Umbrella term for the range of alcohol teratogenesis, including FAS. Ten to 20% of cases of mild ID may be caused by maternal alcohol use. Risk factors: overall alcohol consumption, bingeing, other drug use (including smoking), genetic susceptibility, and low socio-economic status. May be due to the effects of alcohol on NMDA receptors, which may alter cell proliferation. Clinical features—postnatal signs of alcohol withdrawal (irritability, hypotonia, tremors, seizures); microcephaly; abnormal facial features (small eye fissures, epicanthic folds, short palpebral fissure, small maxillae and mandibles, underdeveloped philtrum, cleft palate, thin upper lip); growth deficits (small overall length, joint deformities); CNS features [high incidence of mild ID, associated behavioural problems (hyperactivity, sleep problems), optic nerve hypoplasia (poor visual acuity), hearing loss, receptive and expressive language deficits]; other physical abnormalities (atrial septal defect, VSD, renal hypoplasia, bladder diverticuli). Iodine deficiency disease Worldwide, the most common cause of severe intellectual impairment and largely forgotten in the West by virtue of good diet and iodized table salt. Important cause of ID, particularly because of its treatability. Mainly found in large areas of Asia, Africa, and South America. Congenital hypothyroidism A treatable cause of mental and growth retardation due to loss of thyroid function; incidence 1:4000, but now screened for neonatally and treated early with levothyroxine. If untreated, leads to the typical clinical picture of lethargy, difficulty feeding, constipation, macroglossia, and umbilical

hernia. Secondary to other toxins For example, cocaine, lead, bilirubin, coumarin anticoagulants, phenytoin. Secondary to infective agents ToRCH (Toxoplasmosis, Rubella, Cytomegalovirus, and Herpes simplex virus), syphilis (*Treponema pallidum*), HIV, and other causes of meningitis and encephalitis. Hypoxic damage Secondary to placental insufficiency, pre-eclampsia, birth trauma, severe prematurity, 'small-for-dates' babies (fetal growth retardation), or multiple pregnancy. Central nervous system and skull developmental abnormalities Micro- and macrocephalies, spina bifida, hydrocephalus, craniostenosis, callosal agenesis, lissencephalies, holoprosencephalies.

Disorders of unknown aetiology Disorders of unknown aetiology This includes a broad range of disorders associated with ID, but for which a clear aetiology is as yet undetermined, e.g. cerebral palsies, epilepsy, ASD (E Autism, p. 822), childhood disintegrative disorders, and other clearly defined syndromes with a suspected, but not yet proved, genetic basis (e.g. Cornelia de Lange). Disintegrative disorder Clinical features—characterized by normal development until the age of 4yrs, followed by profound regression with disintegration of behaviour, loss of acquired language and other skills, impaired social relationships, and stereotypies. Aetiology—unknown, but may follow minor illness or viral encephalitis (e.g. measles). Also thought to have a genetic basis, but no genes identified. Prevalence—1 in 100,000. Prognosis—poor, with development of severe ID. Cornelia de Lange syndrome (Brachmann de Lange syndrome) Usually IQ is below 60 (range 30–86); prevalence 1:60,000; mode of inheritance unknown (possibly autosomal dominant), but >99% sporadic. Clinical features—hypertrichosis (hirsutism, synophrys, long eyelashes), facial features (depressed nasal bridge, eye abnormalities, prominent philtrum, thin lips, downturned mouth, anteverted nostrils, bluish tinge around the eyes/nose/mouth, widely spaced teeth, high-arched palate, low-set ears, micrognathia, short neck), limb deformities (especially upper limbs), crypt orchidism/hypoplastic genitals (♂), small umbilicus, low-pitched cry, small nipples. Associated with GI problems, congenital heart defects, visual and hearing problems, skin problems, epilepsy, and death in infancy. Behavioural problems—expressive language deficits, feeding difficulties, sleep disturbance, self-injury, temper tantrums, mood disorders, and autistic features.

820 Chapter 17 Intellectual disability Pervasive developmental disorders Pervasive developmental disorders (PDDs) are a group of lifelong developmental disorders characterized by a triad of: abnormal reciprocal social interaction; communication and language impairment; and a restricted, stereotyped, and repetitive repertoire of interests and activities. In DSM-5 (and ICD-11), the single term 'autism spectrum disorder' is used to describe the range of diagnoses classified as PDDs in ICD-10. ICD-10 currently differentiates: • Autism and atypical autism (E Autism, p. 822). • Rett's syndrome. • Childhood disintegrative disorder. • Asperger's syndrome. • Pervasive developmental disorder not otherwise specified (PDD-NOS). Patients with PDD show either a lack of normal development of skills or a loss of already acquired skills. There is gender bias, with ♂ > ♀ predominance in all syndromes, except Rett's syndrome (♀ predominance). Prevalence of PDD ranges from 10 to 20 cases per 10,000 individuals. Asperger's syndrome⁶ Essence A syndrome first described by Hans Asperger in 1944, but only eponymously named in 1981 by Lorna Wing. Described by Baron-Cohen as 'the extreme male brain', with 'mind blindness'. Characterized by severe persistent impairment in reciprocal social interactions, repetitive behaviour patterns, and restricted interests. IQ and language are normal or, in some cases, superior. Children with Asperger's syndrome may have more striking autistic features before the age of 5 but later develop 'normally' in most spheres, excepting social behaviour. Social deficits commonly manifest in adolescence or early adulthood when the individual experiences difficulty with intimate

relationships. Psychiatric comorbidity is high, with depression and anxiety the most common. Bipolar affective disorder and schizophrenia are more common than in the general population. Mild motor clumsiness (ICD-10) and a family history of autism may be present. Epidemiology ♂ predominance. Prevalence may be as high as 1 in 300, as Asperger's syndrome is almost certainly under-recognized. Rett's syndrome (E X-linked dominant syndromes, p. 814.) Childhood disintegrative disorder (CDD) Rare, occurring in fewer than 5 in 10,000 children. ♂ predominance. There is normal development for 2–3yrs, followed by a loss of acquired motor, language, and social skills between the ages of 3 and 4yrs. Stereotypies and compulsions are common. Cause is unknown, and prognosis is poor. 6 Blasic JC (2008) Pervasive developmental disorder: Asperger syndrome. M <https://emedicine.medscape.com/article/912296-overview> [accessed 12 July 2018]. 7 Bernstein BE (2007) Pervasive developmental disorder: childhood disintegrative disorder. M <https://emedicine.medscape.com/article/914683-overview> [accessed 12 July 2018].

Pervasive developmental disorders Pervasive developmental disorder not otherwise specified Also termed 'atypical autism', PDD-NOS is relatively common and encompasses subthreshold cases where there are impairments of social interaction, communication, and/or stereotyped behaviour patterns or interest, but where full criteria for other PDDs are not met.

822 Chapter 17 Intellectual disability Autism Autism was first described by Maudsley in 1867 and named 'infantile autism' by Leo Kanner in 1943. It is a syndrome that has engendered controversy in terms of its definition, relationship to other syndromes (e.g. schizophrenia), and aetiology. 8 Autism is characterized by the same triad of symptoms (E Pervasive developmental disorders, p. 820) as the core symptoms of PDD: abnormal reciprocal social interaction; communication and language impairment; and a restricted, stereotyped, and repetitive repertoire of interests and activities, in the presence of developmental delay. If this triad of impairments is evident without developmental delay, the diagnosis of 'Asperger's syndrome' or 'ASD' may be given instead. Eighty per cent of patients with autism have mild to moderate ID. In general terms, 1–2% of those with autism have a 'normal' life; 5–20% have a 'borderline' prognosis (i.e. varying degrees of independence), but 70% are totally dependent upon support. Epidemiology The onset of symptoms is typically before the age of 3. ♂:♀ = 3–4:1. Prevalence is 5–10 per 1000 individuals. Aetiology The cause is unknown, but a number of hypotheses exist: genetic (in Down's syndrome and fragile X); obstetric complications; toxic agents; pre/postnatal infections (with maternal rubella); autoimmune; and association with neurological disorders (e.g. TSC). Pathophysiology MRI—some have ↓ brain size; ↓ lateral and fourth ventricles; frontal lobe and cerebellar abnormalities. Pathology Abnormal Purkinje cells in the cerebellar vermis; abnormal limbic architecture. Biochemistry One-third have ↓ serum 5-HT; some have ↓ β-endorphin immunoreactivity. Clinical features • Abnormal social relatedness: impaired non-verbal behaviour; poor eye contact; impaired mentation; failure to develop peer relationships; reduced interest in shared enjoyment; lack of social or emotional reciprocity and empathy; attachment to unusual objects. • Abnormal communication or play: delay or lack of spoken language; difficulty in initiating or sustaining conversation; stereotyped and repetitive (or idiosyncratic) language; mixing of pronouns; lack of developmentally appropriate fantasy, symbolic, or social play. • Restricted interests or activities: encompassing preoccupations and interests; adherence to non-functional routines or rituals; resistance to change; stereotypies and motor mannerisms (e.g. hand or finger flapping or body rocking); preoccupation with parts of objects. • Neurological features: seizures; motor tics; ↓ head circumference; abnormal gaze monitoring; ambidexterity. • Physiological features: unusually intense sensory responsiveness (e.g.

to bright lights, loud noise, rough textures); absence of typical response to pain or injury; abnormal temperature regulation; i paediatric illnesses. 8 Brasic JR (2008) Pervasive developmental disorder: autism. M <https://emedicine.medscape.com/article/912781-overview> [accessed 12 July 2018].

Autism • Behavioural problems: irritability; temper tantrums; self-injury; hyperactivity; aggression. • ‘Savants’: a minority may have ‘islands of precocity’ against a background of ID (i.e. isolated abilities, e.g. incredible memory or arithmetic skills). Differential diagnosis Other PDDs; childhood schizophrenia; ID; language disorders; neurological disorders; sensory impairment (deafness or blindness); OCD; psychosocial deprivation. Assessment9 • A multidisciplinary approach is required, involving psychiatrists, psychologists, paediatricians, neurologists, speech therapists, occupational therapists, social workers, and nursing staff. • Full clinical evaluation, including physical and mental state, as well as specific developmental, psychometric, behavioural, and educational assessments, including clinical observation in different settings. • Rating scales—Autism Diagnostic Interview–Revised (ADI-R); Autism Diagnostic Observation Schedule (ADOS-G); Diagnostic Interview for Social and Communication Disorders (DISCO). Treatment strategies9 • STRUCTURE, ROUTINE, PREDICTABILITY. • Aids to improve communication: symbol dictionaries, picture boards, social stories. • Educational and vocational interventions: special vs mainstream. • Behavioural interventions: includes behaviour modification, social skills training, and CBT methods. • Family interventions: education, support, advocacy. • Speech and language therapy; OT; physiotherapy; dietary advice, etc. • Sleep management. • Pharmacotherapy: symptom management, e.g. short-term antipsychotics for stereotypies (reviewed regularly and only under expert guidance10); SSRIs for compulsive and self-harming behaviours and depression/ anxiety. Treat medical conditions (e.g. epilepsy, GI tract problems). 9 Scottish Intercollegiate Guidelines Network (2016) Assessment, diagnosis, and interventions for autism spectrum disorders. SIGN 145. M <http://www.sign.ac.uk/assets/sign145.pdf> [accessed 12 July 2018]. 10 National Institute for Health and Care Excellence (2012, updated 2016) Autism spectrum disorder in adults: diagnosis and management. Clinical guideline [CG142]. M <https://www.nice.org.uk/Guidance/CG142> [accessed 12 July 2018].

824 Chapter 17 Intellectual disability Epilepsy and intellectual disability Epilepsy is significantly more common in people with ID than in the general population. The prevalence of epilepsy is 740% in the hospitalized ID population and is higher in severe ID (30–50%) than in mild ID (15–20%). It may begin at any age; presentations may change over time, and multiple forms may occur in the same individual. Diagnosis History and examination May be difficult to obtain accurate information, often relying on third-party accounts (home video may be useful). Try to exclude other differential diagnoses (e.g. infection, trauma, hypoglycaemia, hyperventilation, withdrawal from drugs or alcohol, over-sedation, localizing signs of intracranial pathology, evidence of movement disorders). Conduct an MSE, focusing on observed behaviours. Identification of any stressors (especially if anxiety-provoking). Investigations Baseline laboratory tests—FBC, U&Es, LFTs, glucose. Consider EEG and CT/MRI (in complex cases video-EEG monitoring may be useful), PET, or SPECT (to detect areas of hypometabolism). Co-occurrence • Epilepsy is common in patients with ID of various causes, e.g. Down’s syndrome (5–10%), fragile X (25%), Angelman syndrome (90%), Rett’s syndrome (90%). This may be due to shared aetiologies, such as alterations in neuronal development and function, or co-associated brain lesions (haemorrhage, ischaemia, neoplasm, vascular malformation). • Frequent epileptic seizures may lead to (or worsen) permanent loss of

intellectual functioning (e.g. 'acquired epileptic aphasia'/Landau-Kleffner syndrome, progressive partial epilepsies such as epilepsia partialis Kozhevnikov or Rasmussen syndrome type 2), emphasizing the need for early diagnosis and treatment to prevent often fatal progression. Epilepsy syndromes in infancy and childhood Infancy Early infantile epileptic encephalopathy—due to congenital or acquired abnormal cortical development; early myoclonic epileptic encephalopathy, possibly due to metabolic disorders; infantile spasms/West syndrome¹¹ due to intrauterine infections (toxoplasmosis, CMV, rubella), Down's syndrome, TSC, progressive degenerative disorders, or intracranial tumours; severe myoclonic epilepsy. Childhood—a variety of other myoclonic epilepsy syndromes are recognized: Lennox-Gastaut syndrome, myoclonic-astatic epilepsy (Doose syndrome), progressive myoclonus epilepsies (Baltic or Lafora disease), Northern epilepsy. ¹¹ West syndrome is the triad of infantile spasms, mental retardation, and hypsarrhythmia (characteristic EEG finding of chaotic intermixed high-voltage slow waves and diffuse asynchronous spikes).

Epilepsy and intellectual disability Treatment Note: practice varies geographically—in some areas, the lead is taken by neurologists, and in other areas, by ID psychiatrists and/or epilepsy specialist nurses. Choice of treatment will depend upon a number of factors and should be personalized to the patient. It should take into account the type of epilepsy syndrome, possible drug interactions, and side effects. International Association of the Scientific Study of Intellectual Disability guidelines¹² Collation of evidence for different treatments of epilepsy in ID: • Generalized seizures—sodium valproate, lamotrigine. • Partial seizures—valproate, carbamazepine, lamotrigine. Points to note • Behavioural problems may be associated with antiepileptic drugs and may be more common in patients with brain injury or ID (e.g. phenobarbital, primidone, BDZs, vigabatrin). • Communication difficulties may make assessment of side effects more difficult. • For intractable epilepsy, neurosurgery is an option, and it should not be excluded on the basis that the person has ID. Prognosis There is a wide variation in outcome; however, up to 70% of patients with ID can achieve good control of their epilepsy, without major side effects. Common pitfalls • Diagnostic over-shadowing, explaining new (epileptic) symptoms as being 'only' due to the ID. • Epilepsy may be misdiagnosed in patients with ID, particularly when there is a history of sudden unexplained aggression, self-mutilation, and other 'bizarre' behaviours, including abnormal or stereotyped movements, fixed staring, rapid eye blinking, exaggerated startle reflex, attention deficits, or unexplained intermittent lethargy. (If antiepileptic medication has been previously prescribed for these kinds of presentation, consider careful withdrawal, with close monitoring.) • Non-epileptic (pseudo-) seizure disorder can also occur in patients with epilepsy (compare with non-cardiac chest pain in patients with angina). • Epilepsy-related behaviours may also be confused for psychiatric problems, e.g. hallucinations in simple (somatosensory) partial seizures; psychosis-like episodes during complex partial seizures (especially temporal or frontal lobe); or post-ictal confusion. ¹² International Association for Scientific Study of Intellectual Impairment (2001) Clinical guidelines for the management of epilepsy in people with an intellectual disability. Seizure 10:401–9.

826 Chapter 17 Intellectual disability Psychiatric comorbidity in intellectual disability In the assessment of patients with ID, it is important to always consider comorbid psychiatric illnesses, as they are both common and treatable. Psychiatric illness is often missed in the ID population, because of diagnostic over-shadowing (symptoms of mental illness mistakenly attributed to the ID). The diagnostic criteria for people with ID (DC-LD) were published in 2001 by the Royal College of Psychiatrists as an aid to the diagnosis of mental illness in the ID population.¹³ Schizophrenia 73

times more common than in the general population. Age of onset tends to be earlier (mean 23yrs). More commonly associated with epilepsy, negative symptoms of schizophrenia, and impairment of episodic memory.¹⁴ In severe ID, there may be unexplained aggression, bizarre behaviours, mood lability, or mannerisms and stereotypies. Bipolar affective disorder Prevalence is estimated to be greater than the general population (2–12%), with difficulty in making the diagnosis in severe ID. Symptom ‘equivalents’ may include: hyperactivity, wandering, mutism, and temper tantrums. Depressive disorder Commonly missed, as a quiet and withdrawn person may not be a focus of clinical attention. Biological features tend to be more marked, with diurnal variations. Suicidal thoughts and acts may occur in borderline to moderate ID but are less frequent in severe ID. Other causes of mood disturbance (e.g. perimenstrual disorders) should also be considered.

Other disorders Anxiety disorders May be difficult to distinguish from depression, except where there are situational features. OCD Reported to be more prevalent in ID. Differential diagnosis: ritualistic behaviours, tic disorders, behavioural manifestations of autism/ Asperger’s disorder. ADHD Often a prominent feature in children with ID (up to 20%). Stimulants may help in mild ID with clear symptoms, but have no clear efficacy in severe to profound ID. Personality disorder Difficult to define in the ID population, but prevalence is estimated in 720% of mild to moderate ID patients who are inpatients. ¹³ Royal College of Psychiatrists (2001) DC-LD (Diagnostic Criteria for Psychiatric Disorders for Use with Adults with Learning Disabilities/Mental Retardation). London: Gaskell. ¹⁴ Doody GA, Johnstone EC, Sanderson TL, Owens DGC, Muir WJ (1998) Prolapsed schizophrenia revisited: schizophrenia in people with mild learning disability. *Br J Psychiatry* 173:145–53.

Psychiatric comorbidity in intellectual disability

828 Chapter 17 Intellectual disability Behavioural disorders and ‘challenging’ behaviour

Behavioural disorders are over-represented in ID populations, ranging from minor antisocial behaviours to seriously aggressive outbursts. Prevalence estimates are 7% of the ID population: 14% for inpatients (especially 25- to 29-yr olds) and 5% for those in the community (especially 15- to 19-yr olds). Studies of behavioural disorders in the ID population identify six relatively consistent groupings of pathological behaviours,¹⁵ which create a significant burden for parents/carers:

- Aggression–antisocial:
- Antisocial behaviours—shouting, screaming, general noisiness; anal poking/faecal smearing (? secondary to constipation); self-induced vomiting/choking; stealing.
- Aggressive outbursts—against persons or property.
- Severe physical violence—rare.
- Self-injurious behaviour—skin picking, eye gouging, head banging, face beating (more common in severe/profound ID; prevalence 10% overall, 1–2% most severe).
- Social withdrawal.
- Stereotypic behaviours (some of which may be self-injurious).
- Hyperactive disruptive behaviours.
- Repetitive communication disturbance.
- Anxiety/fearfulness.

When these behaviours are particularly severe, they are often termed ‘challenging’ (see Box 17.4).

Associated factors

- Cognitive functioning—severe intellectual impairment, poor/absent language ability, poor social comprehension.
- Temperament—particularly high emotionality, activity, poor sociability.
- Physical problems—e.g. epilepsy, cerebral palsy, cardiac problems, GI problems, visual/hearing impairment.

¹⁵ Einfeld SL, Aman M (1995) Issues in the taxonomy of psychopathology in mental retardation. *J Autism Dev Disord* 25:143–67.

Box 17.4 Definition of challenging behaviour (Royal College of Psychiatrists) ‘Behaviour of such an intensity frequency or duration as to threaten the quality of life and/or physical safety of the individual or others, and is likely to lead to responses that are restrictive, aversive or result in exclusion.’ Royal College of

Psychiatrists. Challenging Behaviour: A Unified Approach. College Report CR144: Royal College of Psychiatrists, British Psychological Society and Royal College of Speech and Language Therapists; 2007 [M Accessed: 12 Jul 2018]: <https://www.rcpsych.ac.uk/usefulresources/publications/collegereports/cr/cr144.aspx>

Behavioural disorders and 'challenging' behaviour • Medication—psychotropic drugs may produce or mask cognitive, behavioural, or emotional problems. Sometimes a 'drug holiday' may be helpful to assess how medication contributes to the presentation. • Psychological factors—e.g. food, drink, pain. • Communication difficulties—frustration due to inability to utilize normal forms of communication. • Adverse experiences—common to the general population and also particular to the ID population, e.g. experience of institutions, social rejection, neglect, and emotional, physical, or sexual abuse. • Environmental factors—living conditions, stability and continuity of day- to-day activities (a common precipitant is multiple short-term residential placements, with multiple changes in care staff). The quality of the care environment may be directly responsible for behavioural problems, and assessment should include factors such as social relationships, specific environmental stressors, consistency of care, and lack of stimulation. • Comorbidity—psychiatric disorders may complicate the presentation of behavioural problems, e.g. ADHD (E Attention-deficit/hyperactivity disorder 1: overview, p. 668); CD/ODD (E Conduct disorders, p. 664; E Oppositional defiant disorder, p. 666); tic disorders (E Tic disorders, p. 676); anxiety disorders (E The common neuroses, p. 362)—fears/phobias, separation anxiety (E Separation anxiety disorder, p. 684), PTSD (E Post-traumatic stress disorder 1, p. 402), OCD (E Obsessive-compulsive disorder 1, p. 384); depressive disorder (E Diagnosis 3: other clinical presentations and differential, p. 252); bipolar disorder (E Bipolar (affective) disorder 1, p. 328); PDDs (E Pervasive developmental disorders, p. 820). Identification and appropriate treatment may significantly improve behavioural problems. Behavioural phenotypes Many genetic causes of ID are associated with characteristic patterns of behaviour. Recognizing these 'behavioural repertoires' may help in diagnosis and management and forms the basis for ongoing research into the genetic basis of some behavioural problems. Examples include: Down's syndrome (oppositional, conduct, and ADHD); fragile X syndrome (autism, ADHD, stereotypies, e.g. hand flapping); Lesch-Nyhan syndrome (self-mutilation); PWS (OCD, multiple impulsive behaviour disorder, e.g. hyperphagia, aggression, skin picking); Smith-Magenis syndrome (severe ADHD, stereotypies— 'self-hugging', severe self-injurious behaviours, insomnia); Williams syndrome ('pseudomature' language ability in some; initially affectionate and engaging; later anxious, hyperactive, and uncooperative).

830 Chapter 17 Intellectual disability Management of behavioural disorders At all stages in assessment and management, it is essential to involve parents, carers, and other allied professionals (e.g. teachers), both as sources of information and in implementing any proposed interventions. Assessment • Exclusion of psychiatric disorder. • Exclusion of physical disorder and assessment of general health. • Assessment of physical impairments (vision, hearing, etc.). • Assessment of communication difficulties (including formal speech and language assessment). • Assessment of specific cognitive impairments (including formal psychological testing). • Identification of environmental and social factors. • Functional assessment of behaviour (including description of behaviour, situations, consequences, and reinforcers).¹⁶ Management Following assessment, specific factors should be addressed—psychiatric/ physical causes, reduction of stimuli/reinforcers, and modification of environmental factors. Approaches may involve: • Educational interventions—both for families/carers (to improve understanding) and for patients (to

ensure educational needs are being appropriately met in a suitable setting). • Social interventions—to address unmet needs at home, with family/ carers, or to widen access to other services or facilities (to provide opportunities for social interaction and improve support networks). • Facilitating communication of needs—addressing impairments of hearing, vision, and language (including use of pictures, sign language, and electronic speech devices). • Behaviour support plan—identifying proactive strategies to improve quality of life, adaptations and strategies to change behaviour, preventative strategies to prevent distress, and reactive strategies to deal with challenging behaviour.¹⁷ • Cognitive approaches—at an appropriate level for the degree of cognitive impairment and language abilities—may range from counselling on specific issues to simple imitation of relaxation/breathing techniques. • Pharmacotherapy—treatment for specific comorbid conditions (e.g. ADHD—stimulants; OCD—SSRIs, antidepressant treatment; tic disorders—antipsychotics; epilepsy—anticonvulsants). Medication to treat challenging behaviours should only be used if the risk of harm to the patient or others is high or if other interventions have failed. Other 16 National Institute for Health and Care Excellence (2015) Challenging behaviour and learning disabilities: prevention and interventions for people with learning disabilities whose behaviour challenges. M <https://www.nice.org.uk/guidance/NG11> [accessed 12 July 2018]. 17 National Institute for Health and Care Excellence (2015) Challenging behaviour and learning disabilities: prevention and interventions for people with learning disabilities whose behaviour challenges. M <https://www.nice.org.uk/guidance/NG11> [accessed 12 July 2018].

Management of behavioural disorders interventions should be continued, where possible, and medication reviewed on a regular basis. Sometimes a trial of antipsychotic treatment may be useful for serious aggression, hyperactivity, or stereotypies (caution in epilepsy; i risk of EPSEs). Other options for aggression, agitation, or self-injurious behaviours (mainly empirical evidence): anticonvulsants, lithium, β -blockers, buspirone. For self-injurious behaviours alone, there is some evidence for opiate antagonists (e.g. naltrexone). • Physical interventions (i.e. restraint)—from splints and headgear to isolation (to protect the individual and others from injury/damage to property). This should only be done as a last resort to protect the individual or others. Any intervention should be closely monitored to ensure compliance, acceptability, and therapeutic response. In the case of medication, side effects should be minimized, and if treatment is deemed ineffective, drugs should be carefully withdrawn (to avoid secondary problems).

832 Chapter 17 Intellectual disability Forensic intellectual disability The rate of offending in people with ID is consistently higher than that of the general population, with some studies estimating up to around 30% of people with ID come into contact with the criminal justice system.¹⁸ As well as a higher arrest rate, persons with ID have a higher prosecution rate, related to a greater tendency to plead guilty and being less likely to plea bargain. Evidence also shows that people with a more severe ID are less likely to offend than those with mild or moderate disability. Specific types of offence • Crimes of aggression are more common in people with ID. Aggressive behaviour in patients with ID has a wide differential (E Behavioural disorders and 'challenging' behaviour, p. 828), including aggression associated with ictal or post-ictal states in patients with epilepsy and in association with ASD. • A higher rate of sexual offending has been reported in people with ID. The reasons behind this are complex and not particularly well understood. This may relate to poor sexual knowledge, together with an attempt to fulfil normal sexual urges. Sexual Offender Treatment Programmes (SOTPs) can be adapted for those with ID. Use of libido suppressants is possible but remains controversial. • Arson is another offence in which people with ID are over-

represented. There appears to be a split in the reasons behind this, with some incidents being viewed as a 'cry for help' and others being due to a fascination with fire. A CBT programme has been developed in certain forensic units in England which is specifically for people charged with arson. Assessment of people with forensic issues and intellectual disorder • Given that offending behaviour in people with ID is seen as part of the challenging behaviour spectrum, assessment of this should be used as a basis for assessment of people in the forensic setting. This involves early identification of people with a higher risk of going on to develop offending behaviour, including those with environmental risk factors. • The process should be flexible, dependent on the person's level of disability and functioning, as well as their needs. As part of the assessment, there should be a functional assessment of the behaviour, looking for any particular triggers which are potentially reversible. 18 Loucks N (2007) The prevalence and associated needs of offenders with learning difficulties and learning disabilities. London: Prison Reform Trust. M <http://www.prisonreformtrust.org.uk/uploads/documents/NOKNL.pdf> [accessed 12 July 2018].

Forensic intellectual disability Criminal justice and intellectual disability • The criminal justice system can seem overwhelming to someone with an ID, and it is important they have appropriate support at all stages of the process (E Appropriate adults, p. 754). The National Appropriate Adult Scheme¹⁹ aims to ensure that all vulnerable people have access to a trained person who can ensure that the person's rights are being respected and make sure they understand when they are being interviewed by the police. • If a person with an ID is charged and has to go to court, they should be assessed for their fitness to plead (E Fitness to plead 1: assessment, p. 774). Even if they are fit to plead, a mental health disposal or treatment order may be more appropriate and they can be cared for in a secure unit, if required. • People with IDs are, by their very nature, more vulnerable than some others, which can lead to bullying and intimidation within the prison system. They may also struggle with communication within the prison, and it is important that people working with them are made aware of this. • A number of treatment programmes developed within the prison system can be adapted for people with ID, meaning they have the same access to treatment as other prisoners. 19 For further information, see: M <http://www.appropriateadult.org.uk> (England, Wales, and NI); M <http://www.gov.scot/resource/doc/1099/0053903.pdf> (Scotland) [both accessed 12 July 2018].

834 Chapter 17 Intellectual disability Transition periods Adolescence This may be a difficult transition period; issues that may require attention include: • Engaging with adult services Loss of the additional support provided by supported mainstream or special schools may lead to problems if there is not a smooth transition to adult services. Where appropriate (or available), this may include moving to social educational/day centres. Some countries have specific legislation to ensure that needs are identified early. • Social/economic independence • Employment—depending on the level of disability, this may be in sheltered employment, workshops, or supported open employment. Despite changing attitudes, there are considerable barriers to finding work in the open job market, although, for some, this may be worth pursuing. • Living arrangements—loss of additional social supports may actually increase the burden of care shouldered by the family. For some, the wish for independence or the lack of family support may be best met with small group homes where support may be tailored to individual needs. • Sexual relationships Societal views may find it difficult to accept the fact that people with ID have normal sexual desires, which can be more of a problem for families/carers than the individuals themselves. Nonetheless, issues raised by appropriate sexual relationships will include consideration of contraception, understanding of

the responsibilities of parenthood, and issues of commitment and marriage. Many people, particularly with mild ID, are capable of being successful parents and provide a stable environment for children, with appropriate support. Policy and practice guidelines will often exist on this contentious topic, e.g. the 'Making Choices Keeping Safe' policy in Lothian.²⁰

Later adulthood

- **Changing health needs** With increasing age, health needs may go unrecognized and there may be failure to access services. Patients with ID may lack capacity to consent to necessary medical treatment, but this should not be allowed to prevent appropriate treatment.
- **Changing mental health needs** These may relate to changing symptomatology over time, altered tolerance to medication, and additional specific age-related cognitive impairment (e.g. due to chronic intractable epilepsy or early-onset Alzheimer's disease in Down's syndrome).

²⁰ Making Choices Keeping Safe (MCKS). Guidelines for those caring for people with learning disabilities around relationships and sexual health. The latest guidelines (May 2016): <https://services.nhslothian.scot/LearningDisabilities/Pages/MakingChoicesKeepingSafe.aspx> [accessed 19 January 2019].

Transition periods

- **Ageing carers** The ability of carers to continue to provide the same level of care for their children ought to be considered before a crisis is reached. This requires an ongoing assessment of the patient's needs and the carer's abilities. Increasing reliance on carers may lead to social isolation for the patient, and it is prudent to raise the issue of planning for the future at an early stage. Death of carers may produce multiple simultaneous difficulties when a patient with ID must cope with bereavement, loss of a familiar home setting, and adjustment to new carers and living with other individuals in a group setting.

836 Chapter 17 Intellectual disability Family issues Having a child with ID is a major, and often unexpected, blow to any family. Individual responses vary, but the majority of parents adapt well to the situation and show remarkable resilience and resourcefulness. Depression is quite common in parents and should not be overlooked. Important positive factors include having a good relationship with their partner and the support of relatives and friends. Needs and priorities will vary over time and should be identified early and addressed collaboratively, with the involvement of parents and other carers in any key decisions (E Needs and priorities, p. 837).

Early impact

Prenatal diagnostic screening can place parents in the unexpected position of having to make difficult choices, even before the birth of their child. Advice and counselling are a necessary and important part of the screening process and should not be ignored, even when testing is regarded as 'routine'. The mistaken assumption that screening 'guarantees' a healthy child may lead to even greater feelings of disappointment and anger, magnified further by anxious times after the birth, with a baby in a special care unit. Although some conditions can be diagnosed at birth, often parents only realize there is a problem when their child fails to reach developmental milestones or develops seizures after an apparently 'normal' infancy. Often the response is one of bereavement (E Normal and abnormal grief, p. 400) or guilt, and parents may need support to 'work through' their feelings. The importance of diagnosis A clear diagnosis is essential and may greatly relieve the anxieties of many parents who may blame themselves for their child's problems. It may allow access to specific supports, including parent groups and support organizations. These can provide valuable support and education and help answer the many questions which parents have (e.g. usual course, associated problems, prognosis). For inherited conditions, the issue of further genetic counselling/testing of family members needs to be addressed. Provision of clear information allows individuals to make informed decisions about being tested and to weigh the risks of having other

affected children. The effect on other family members Although it was previously thought that having a child with ID impacted adversely on other unaffected siblings (often leading to the removal of the child from the family), there is little evidence that this is the case and worries about long-term damage appear unfounded. In fact, brothers and sisters of individuals with ID often appear to be drawn to the caring professions and many end up working as doctors, nurses, teachers, or providing support for children with special needs. Grandparents may be a useful supportive resource for parents but may also need to come to terms with their own feelings of having a disabled grandchild.

Family issues The 'burden of care' For carers, informal support may actually be more valuable than formal (professional) support. Frequent appointments or regular home visits may be more disruptive than helpful. Developmental delay brings with it associated problems (e.g. longer time until the child can walk, achieve continence, acquire language/communication skills, establish a normal sleep pattern). The social, financial, and psychological impact on carers should be acknowledged, and appropriate help and support provided. For infants and children, schooling may be both a benefit (in terms of learning social skills, support/respite for parents, and close contact with teachers/other parents) and a burden (particularly if necessary specialist schooling is not locally available). Transitional periods (e.g. adolescence/early adulthood) are accompanied by parental anxieties, as well as changes in how needs are met (E Transition periods, p. 834). Advance planning will go some way to alleviate i carer stress. Carers may also be concerned about what will happen to their child when they are no longer able to care for them, and an open discussion of these issues, with provisional planning, may help avert crises. Needs and priorities

- Early, accurate diagnosis.
- Informative genetic advice to parents and other family members.
- Access to high-quality primary (and secondary) healthcare.
- Advice and access to appropriate help and support (practical help, financial assistance, social and educational needs).
- Help and advice with any communication problems (communication aids, learning of sign language).
- Consideration of the needs of carers (education, support groups, respite care).
- Provision of specialist and domiciliary help with specific behavioural problems.
- 'Safety net' of open access to i support when necessary.
- Acknowledgement that needs will change over time (and planning for this; E Transition periods, p. 834).

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