1: Definitions, classifications and epidemiology of intellectual disability (ID), *Sally Ann Cooper*

2: Clinical Assessment Including Bedside Diagnosis, *Sabyasachi Bhaumik, Regi Alexander, Shweta Gangavati, Rohit Gumber, and Samuel Tromans*

3: Intellectual disability - concepts, aetiology and genetics, *Satish C. Girimaji, Salah Basheer, Asit Biswas, and Satheeshkumar Gangadharan*

4: Children and Adolescents with Intellectual Disability, *Tom Berney and Mark Lovell*

5: Measuring outcomes including use of rating scales and instruments in people with intellectual disability, *Peter Tyrer, Freya Tyrer, Marissa Hanney, and Stephen Tyrer*

6: Types of mental disorders in people with intellectual disability, *Sally Ann Cooper*

7: Autism Spectrum Disorder, *Mary Barrett and Traolach S. Brugha*

8: Attention Deficit Hyperactivity Disorder (ADHD) in people with intellectual disability, *Kiriakos Xenitidis, Shazia Zahid, and Caryl Marshall*

9: Dementia and other disorders associated with ageing in people with intellectual disability, *Ande Strydom, Karen Dodd and Nwamaka Uchendu, and Suzanne Wilson*

10: Schizophrenia and related psychoses in people with Intellectual Disability, *Laura Humphries, Dasari Michael, and Angela Hassiotis*

11: Depressive Disorders in People with Intellectual Disability, *Samuel Tromans, Atiq Umar, Jennifer Torr, Regi Alexander, and Sabyasachi Bhaumik*

12: Bipolar Affective Disorder in Intellectual Disability, *Samuel Tromans, Ian Jones, Ignatius Gunaratna, Natalie Orr, and Sabyasachi Bhaumik*

13: Anxiety and related disorders in people with Intellectual Disability, *Sherva Cooray and Avinash Hiremath*

14: The assessment and treatment of personality disorders in people with intellectual and developmental disabilities, *Regi Alexander, Verity Chester, and William R. Lindsay*

15: Alcohol and Substance Misuse and People with Intellectual and / or Developmental Disabilities, *Tobias Buxton, Verity Chester, Bijil Arackal, Divya Devulapally, and Yona Lunsky*

16: Challenging behaviours in people with ID, *Glynis H. Murphy and Peter McGill*

17: Offending Behaviours in People with Intellectual Disabilities, *John L. Taylor, William R. Lindsay, and John Devapriam*

18: Psychological therapies with people who have intellectual disabilities, *Peter E. Langdon, Tawakalitu Kehinde, and Georgina Parkes*

19: Pharmacotherapy for Mental Illness and Behaviours that challenge in People with Intellectual Disabilities, *David Branford, Reena Tharian, Regi Alexander, and Sabyasachi Bhaumik*

20: Physical Health in people with Intellectual disabilities, *Umesh Chauhan, Ken Courtenay, and Matthew Hoghton*

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23: Behaviour Phenotypes, *Anthony Holland*

24: Visual and Hearing Impairments and their Impact on the Mental Health of Adults with Intellectual Disability, *Reza Kiani and Sugato Bhaumik*

25: Family Experiences of Psychiatric Services for their Relative with Intellectual and Developmental Disabilities, *Verity Chester, Neil James, Ian Rogers, Jackie Grace, and Regi Alexande*

26: Current and Future Research Priorities in the Psychiatry of Intellectual Disability, *Rupert Thurston, Samuel Tromans, Verity Chester, Sally-Ann Cooper, Andre Strydorm, Sabyasachi Bhaumik, and Dheeraj Rai*

27: Models of service development and delivery for people with Intellectual Disability, *Sujeet Jaydeokar, John Devapriam, Jane McCarthy, Chaya Kapugama, and Sabyasachi Bhaumik*

28: Safeguarding and ethical practice for people with intellectual disability, *Meera Roy, Ashok Roy, Priyanka Tharian, Ameeta Retzer*

2

Clinical Assessment Including Bedside Diagnosis

Sabyasachi Bhaumik†, Samuel Tromans, Rohit Gumber, and Shweta Gangavati

Introduction

Intellectual disability (ID) is a heterogeneous condition and includes people with wide-ranging abilities (i.e. from those with reasonable verbal skills to those who can only communicate through eye movements, facial expressions, or other simple gestures such as nodding or finger pointing). At the one end of the spectrum, it is possible to have a good verbal interaction especially with those with mild ID. However, at the other end, you may encounter people with ID with no verbal communication and very limited non-verbal expressions. Hence, interviewing someone with ID requires adaptations in line with the person's communication needs.

Many aspects of psychiatric assessment in People with Intellectual Disabilities (PWID) bear similarity to that of the general population. However, there are also considerable differences involving the areas of information gathering, psychiatric interviewing, mental state examination, and the importance of observational skills, especially for non-verbal patients. There is a considerable emphasis on physical examination, as the interface between the physical and mental states often overlap, having great influence of one another. In addition, there are also differences in interviewing techniques involving adults, children, and the older population. One of the key central issues is of how to take a psychiatric history that is meaningful for a non-verbal patient and interpreting one's findings in a systematic way. The symptomatology of psychiatric disorders are varied, but very often these symptoms present in an atypical fashion in those with ID.

A shift from the normative function of that person may indicate underlying mental health problems, physical health problems, environmental issues, or communication difficulties. To illustrate, we provide an example of a patient who presents with the symptoms of early morning wakening; however, on closer questioning and further enquiries you may find out that the person concerned has always been an early riser and tends to get up at around 4:30am. This symptomatology therefore does not fit into part of the symptom cluster for a depressive illness, but rather is a normative function for that person. When you complete your assessment and agree for a plan of interventions, your therapeutic goals should be to return to the baseline function of that person, otherwise there will be disappointments for the high expectations that you may have given to both carers and the person with ID in relation to the outcome of your interventions. So please go through the following sections carefully, bearing in mind that they highlight the key differences between the general interviewing process and the interviewing process for PWID.

This chapter will highlight some of these issues and hopefully help trainees feel more prepared to carry out effective psychiatric assessments in PWID.

Psychiatric interviewing

The main goals of a psychiatric interview in a PWID are to:

- Build rapport with the patient
- Arrive at a diagnosis if possible
- Understand the causation through contextual information

Fundamental to interviewing a PWID, and indeed psychiatric interviews more generally, is establishing a good level of rapport with the patient. A useful tip is to learn about the patient's likes and dislikes prior to the meeting; this can help inform how you can arrange the meeting in a way that is conducive to providing a supportive atmosphere that will cause them minimal distress. For example, many PWID find clinical environments intimidating or even frightening, and thus a natural setting such as the patient's own home, may be preferable. Sometimes PWID may require time to relax or settle into a meeting, and starting by discussing a topic of their interest may help in this regard, rather than immediately delving into the psychiatric interviewing process. The presence of a carer who knows the patient well is also a useful technique for putting the patient at ease.

Whilst arriving at a diagnosis is a central tenet of the psychiatric interview in a PWID, you may often only be able to arrive at a probable working diagnosis initially, with a clear diagnosis often emerging through the process of time and further information gathering. Diagnoses may have physical and psychiatric aspects

† It is with great regret that we report the death of Sabyasachi Bhaumik during the production of this textbook.

to them, and it is important to appreciate the marked effect that common physical health conditions (such as chronic pain) can have on behaviour and mental state. Also, it is essential to be mindful of the possibility of underlying psychological, physical, or sexual abuse, as this is more common in the ID population, and can lead to challenging behaviours that maintain a cycle of abuse (1). In situations where it is impossible to rule out other potential diagnoses, you might be best placed to begin the process of a therapeutic trial based on your priorities. For instance, if you strongly suspect a bipolar disorder but cannot be sure the patient is not suffering with paranoid psychosis, you might use a therapeutic trial. Commencing multiple therapies at the same time, especially medications, is not recommended as you will not know what agent has had the positive effect for the patient.

Contextual information is important, particularly from family carers, who will usually have known the patient for many years and likely be able to provide a wealth of relevant clinical information. In speaking to foster or professional carers, request to speak to those whom have known the patient for longer than six months, in order to improve one's chances of obtaining a meaningful history. Please also be mindful about the influence of the environment in which the patient lives, along with attitudes of care staff. Thus, it is important to make the patient the centre of any assessment approach. Additionally, observing the patient directly can be helpful in situations where you receive conflicting reports from carers.

Stages of the assessment process

Preparation and collection of information

Please ensure that you have gathered and gone through all the pertinent information from the relevant sources prior to the assessment. Occasionally, there may be a need to correspond further with the referrer prior to the assessment, especially if the information provided is perceived to be incomplete. Any further information gap might be filled up during the assessment process itself.

Of course, the assessment itself provides the main opportunity to obtain information relevant to the patient's current mental state and functioning, as well as filling in any informational gaps identified in the preparatory stage. Therefore, please be mindful that information collection during the process should not lead to ignoring the patient's wishes and missing the opportunity to establish rapport. Purely information collection alone is insufficient and one must make every effort to ensure that the assessment experience is a positive and valuable interaction for the patient as well. Thus, endeavouring to put the patient at ease, establishing rapport, and ascertaining the patient's wishes are all vital in any successful consultation. You may wish to consider inviting other healthcare professionals to the assessment, which might enable you to obtain their views and establish a multidisciplinary care plan.

An initial psychiatric assessment for a PWID typically takes between 60 to 90 minutes, though the time required may be more in patients with particularly complex needs. Follow-up clinic assessments for patients already known to services are often around 30 minutes duration, though again one needs to be flexible and individualize their approach according to the specific patient involved.

Using the information

Needless to say, simply collection of the relevant information is insufficient; it needs to be considered and formulated to develop an appropriate management plan, making the assessment a worthwhile endeavour for the patient and that their needs are suitably addressed.

Recording and communicating the information

The information collected from the assessment process needs to be accurately recorded in the patients clinical notes, to provide a reference point for other health professionals in working with the patient in the future. Additionally, these details need to be communicated to the professional who made the original referral to ID services, as well as any other appropriately involved parties.

Capacity to consent

Capacity assessment is essential to decisions made across all medical care, but additional challenges in PWID, pertaining to their ID as well as other factors such as communication impairments, make them an even more prominent part of day-to-day practice. It is imperative that every attempt to communicate the information in as simple terms as possible, via the patients preferred mode of communication.

In making treatment decisions for patients who lack the capacity to consent, ensure that all relevant parties have been consulted with regard to making a best interests decision, including closest relatives, professional carers and members of the multidisciplinary team. Additionally, be mindful that capacity is both decision and time-specific. For example, following antipsychotic treatment for an acute psychotic episode, a patient may have regained capacity to make her decision regarding whether to take this medication upon restoration of their baseline mental state.

It is worth recognizing that, as ID psychiatrists, we have specialist expertise in assessing capacity relative to many of our peers, and that we can support other professionals in making such judgements for PWID.

Sharing information with families and caregivers is an important part of working collaboratively with PWID. However, where the patient has capacity to make the decision about this, informed consent should be obtained. In instances where the patient lacks consent, a best interests decision should be made by the multidisciplinary team managing the patient's care on whether sharing such information is best for the patient. In most such instances, information sharing will likely have a positive effect on the patient's care, as well as empowering families and caregivers; however, in exceptional circumstances such as instances of suspected abuse, information may be withheld in the patient's best interests.

Communication skills

As discussed in the introduction, communication skills vary widely within the ID population, and one must modify their interview approach accordingly. Some patients may be able to communicate verbally on a level not too dissimilar to those in the non-ID population. However, it is important not to assume that relatively good communication skills necessarily indicate comprehension, and to check the patient's level of understanding throughout the assessment process, particularly with regard to any therapeutic decision-making.

It is important to appreciate that you may be seeing the patient at a time where there are concerns pertaining to their mental state and/or functioning, and as such, their communication skills may be similarly affected. Information from carers as well as previous clinical assessments are central to determining if this is the case. A speech and

language therapist may have developed a communication passport for the patient following a detailed assessment, which can contain vital information of how best to approach your assessment.

Patients have different preferred means of communication, and predominantly verbally-based interaction with the patient may be inappropriate in some instances. It is worth considering why the patient prefers to communicate in a particular way, as the underlying basis for such inclinations may lie in an undiagnosed sensory deficit. Some patients respond well to a picture-based approach to communication, particularly with regard to explaining multi-step processes (e.g. visiting the doctor).

History

Developmental history

ID is a developmental disorder, and one of the key distinctions between taking a history from a PWID relative to someone without ID is the level of depth explored with regard to their development. Thus it is essential to have a good working understanding of the major developmental milestones (see Table 2.1). Often patients with suspected or known ID will have had previous assessments conducted during the developmental phase (0–18 years of age), and, where this information is available, the findings and conclusions can be invaluable in determining possible aetiology, characterizing their behavioural phenotype and understanding how their functioning has changed in the intervening period. Certain genetic syndromes associated with ID have characteristic behavioural phenotypes and associated ID severities (see Table 2.3), and thus gaining a thorough understanding of development can support one's clinical suspicions and prompt collaborative assessment with a clinical geneticist specializing in genetic causes of ID.

It is conceivable that a patient may now demonstrate measured intelligence and adaptive functioning consistent with ID, but that this was not always the case for them. Indeed, the definition of ID requires an onset before the age of 18 years (i.e. the developmental period), otherwise their impairments would instead be considered as an adult onset insult to brain. Conversely, if someone has acquired brain injury during the developmental period, he or she will come under the umbrella of ID.

Understanding adaptive functioning

Initially, information on adaptive functioning can be obtained from both patient and carer, as well as other informants. Assessing particular aspects of functioning (e.g. reading and writing) can sometimes be assessed directly as part of the assessment process, though it should be borne in mind that if the patient's mental state is not currently at its baseline level, their functioning will likely be similarly disrupted (see Table 2.2).

It can be invaluable to take a history of a 'typical day' for the patient, from first thing in the morning to last thing at night, where each daily task is discussed in detail with regard to the level of independence and/or degree of support the patient requires. In instances where the patients functioning has changed (e.g. due to psychiatric illness), the discrepancies between current and previous functioning can be ascertained and later used as a means of establishing their subsequent progress post-intervention.

Diagnostic criteria for ID include requirements for both intellectual and adaptive impairments. At present IQ tests are rarely carried out; where it is available, it is important to understand it clearly. However, there is a potential for fluctuation in IQ scores at different phases of life. In the absence of an available IQ score, you may wish to use certain measures of assessment of adaptive functioning, such as ABAS and Vineland adaptive behaviour scale, both of which will give a clear idea as to the patient's approximate mental age. In assessing intelligence, clinicians may choose between standardized measures of IQ, some of which measure overall intellectual ability by measuring two core domains—verbal and non-verbal reasoning skills, while others include additional domains such as information-processing. The most commonly used IQ tests, the Weschler Intelligence Scale for Children, fourth edition (WISC-IV) and the Weschler Adult Intelligence Scale, fourth edition (WAIS-IV), offer both a full-scale IQ (FSIQ) and general abilities index (GAI). The four index scores representing major components of intelligence are verbal comprehension index, perceptual reasoning index, working memory index, and processing speed index. Evidence suggests that the use of full scale IQ in ID diagnostic decision-making, which includes working memory and processing speed, is a more appropriate measure as these impairments are vital components in the efficiency of overall intellectual functioning (2,3). A number of measures for the assessment of adaptive functioning are currently available, the two most commonly used in the United Kingdom are the Adaptive Behaviour Assessment System, second edition (ABAS-II) (4) and the Vineland Adaptive Behaviour Scales, second edition (Vineland-3) (5).

The task of determining whether the individual has impaired social functioning (adaptive behaviour) an IQ less than 70, is not as straightforward as the definition implies. There are a number of problems associated with the tools used. The reliability of IQ tests has been called into question; it is well-known that IQ scores are only accurate to within about five points; however this may be considerably less accurate when applied to people with low IQ. There is also evidence to suggest that there is a lower retest reliability for individuals with low IQ. Even amongst the two most commonly used intelligence scales, there is evidence of significant lack of consistency in the low IQ ranges. Evidence suggests that the Weschler Intelligence Scale for Children, fourth edition (WISC-IV) may give IQ scores of approximately 12 points lower than the Weschler Adult Intelligence Scale, third edition (WAIS-III) (6, 7). The reliability of adaptive behaviour scales has also been called into question. The reliability of self-report measures are especially salient when respondents could have a potential stake in the outcome of the assessment. This may also hold true for third-party respondents where there may be a potential secondary gain. For the test results to be considered valid it is important that the individual gives full effort to the assessment process, and lack of motivation to do so or deliberately underperforming during formal assessment will clearly impact on results. Clinicians should consider and formally assess, where indicated, the person's efforts throughout the assessment. Conversely, in some circumstances it has also been suggested that individuals have a tendency to overestimate the competence and adaptive skills in an effort to appear more capable than they may actually be. The only instrument available that permits self-report is the ABAS-II, however the authors clearly state that they do not recommend relying on self-report for the purposes of ruling in or out a diagnosis of

Table 2.1 Summary of developmental milestones during the first 4 years of life

Average age	Milestone	Red flags
6 weeks	• Smiles • Follows eyes past midline	
4–6 months	• Sits with support • Rolls • Reaches out and grasps objects • Starts babbling	At 6 months if: • No smile • No grasp • Not rolling • Poor head control
6–9 months	• Crawls • Sits without support • Pulls to stand • Transfers objects between hands • Gives toy on request • Turns head to name • Responds to 'bye-bye' • Gestures with babbling • First tooth	At 9 months if: • No response to words • Lack of eye contact or facial expression • No gestures • No passing of toys from hand to hand • Not sitting without support or crawling
7–12 months	• Develops pincer grasp • Plays 'peek-a-boo' • Walks with a hand held • Waves goodbye	At 12 months if: • Unable to pick up small items • Not crawling/ bottom shuffling • Not standing when holding onto furniture • No babbled phrases
12–15 months	• Single words • Listens to stories • Drinks from cup • Builds 2-brick tower	
18 months	• Speaks 6 words • Able to walk up steps • Names pictures • Walks independently • Scribbles • Builds 3-brick tower • To-and-fro scribble	• Uninterested in playing with others • No clear words • Not walking without support • Not able to hold crayon • Unable to stack 2 blocks
2 years	• Kicks/ throws a ball • Runs • Jumps • 2-word sentences • Understands two-word commands (e.g. 'feed teddy') • Builds 6-brick tower • Turns pages • Uses a spoon • Helps with dressing • Circular scribble	At 2 years if: • Has <50 words • Difficulty handling small objects • Unable to climb stairs • No interest in feeding or dressing
3 years	• Stands on one leg momentarily • Asks 'what' and 'who' questions • Uses pronouns (e.g. 'I', 'me') • Understands three-word commands • Eats with fork and spoon • Participates in imaginative play • Draws circle	
4 years	• Hops • Can dress and undress (except for shoe laces) • Asks 'why', 'when' and 'how' questions • Can count up to 20 • Draws person with head, body, and legs	
If there is regression, or loss of a previously developed skill, this should be considered a red flag requiring immediate investigation. Other red flags at any age include poor interaction with others, differences in strength between right and left sides of body, abnormal tones and strong parental concern.		

Adapted from *Oxford Handbook of Clinical Specialities*, 9th edition, Judith Collier, Murray Longmore, Keith Amarakone, p.219, 2013. Source data from *Training in Paediatrics*, Mark Gardiner, Sarah Eisen, Catherine Murphy (eds), 2009. By permission of Oxford University Press.

Table 2.2 Adaptive Functioning Across Levels of Intellectual Disability

	MILD IQ (69–50)	MODERATE IQ (49–35)	SEVERE/PROFOUND IQ (34–0)
Mental Age Equivalence	9yrs–<12yrs	6yrs–<9yrs	3yrs–<6yrs
Proportion	85% of the group	10% of the group	3%–4% of the group
Language Acquisition	Some delay Most achieve the ability to use speech for everyday purposes, hold conversations and engage in clinical interview. Executive speech problems may persist. and interfere with development of independence.	Slow in developing comprehension and use of language. Eventual achievement is limited but variable: from just enough language to communicate basic needs to being able to have simple conversations. May learn to use manual signs to compensate.	Acquire little or no communicative speech in early childhood years but may develop some speech during school-age period.
Expressive Language	Most achieve the ability to use speech for everyday purposes, hold conversations and engage in clinical interview. Executive speech problems may persist and interfere with development of independence.	From just enough language to communicate basic needs to simple conversations with limited vocabulary. May learn to use manual signs to compensate.	Limited to a few words only or absent speech May indicate choice through nodding or pointing.
Comprehension	Reasonable	Limited to simple instructions.	Very limited understanding if any
Non-verbal communication	Good	Limited	Rudimentary
Self-care & Continence	Most achieve full independence in washing, eating, dressing as well as bladder and bowel control.	Can attend to personal care with moderate assistance from carers. Mainly continent.	Achieve elementary skills only. Full support of carers needed. Mainly incontinent.
Independent Living	Full independence in practical and domestic skills possible. May be able to cook simple meals; participate in household chores; operate common household appliances (television, telephone, microwave, washing machine, etc.). May travel independently; do everyday shopping and use money. Regression in skills is common under unusual social or economic stress	Will need supervised living arrangements. Limited mastery of domestic tasks; will require support and assistance. Unlikely to shop or use public transport without support.	Full 24-hour supervision required.
Academic Skills	More likely to have left school without any qualifications; achievements up to approximately the sixth-grade level. May learn to read, write, and do simple maths but can have problems.	More likely to have attended a special school; achievements unlikely beyond the second-grade level. May develop some reading, writing and math skills.	Familiarity with the alphabet and simple counting. Simple sight reading of some words. May learn to copy/write. Simple visuospatial skills.
Adult Work	Capable of work demanding of practical rather than academic skills.	Simple practical work with supervision.	Most not capable of this.
Motor Skills	Normal mobility. Generally without problems with motor dexterity.	Delayed but usually fully mobile.	Frequent musculoskeletal abnormalities. Often severe restriction.
Social & Emotional Development	Some immaturity is present which can make demands of marriage, child-rearing, or fitting in with cultural traditions and expectations difficult.	Interaction may be as usual but difficulties in understanding social conventions may interfere with peer relationships	Maybe very limited. Autism common.
Associated Deficits	Organic aetiology identifiable in only a minority. Minimal sensorimotor impairment. Other deficits as in normal population.	Organic aetiology identifiable in a greater proportion. More frequent sensorimotor impairments with Increase in CNS disorders like epilepsy.	Organic aetiology frequently identifiable. Increased CNS disorders such as epilepsy and sensorimotor deficits including visual and hearing. Impairments.
Autism & other Pervasive Developmental Disorders	Present in varying proportions.	Present in a substantial minority and can impact clinical picture and type of management needed.	Increased prevalence affecting presentation and management.

Table 2.3 Overview of genetic disorders associated with ID

Genetic syndrome	Underlying genetic pathology	Degree of ID	Morphological features	Behavioural phenotype	Physical associations	Psychiatric associations
Angelman syndrome	Lack of maternal contribution to a portion of chromosome 15	Severe	Microcephaly, strabismus, coarse facial features, hypopigmentation of skin and eyes	Hand flapping, frequent laughter, happy demeanour, excitability, attraction towards water	Seizure disorder, ataxia, scoliosis	Speech impairment, sleeping difficulties
Cornelia de Lange syndrome	Can be due to mutations in different genes (e.g. NIBPL, SMC1A, SMC3, HDAC8, RAD21)	Moderate to severe	Microcephaly, unibrow (synophrys), small and upturned nose, low-set ears, large philtrum	Aggression, self-injurious behaviour	Congenital heart disease, cleft palate, hearing impairment, gastro-oesophageal reflux	Autism spectrum disorder or ASD-like traits
Cri du chat syndrome	Deletion of a section of the short arm of chromosome 5	Moderate to severe	Microcephaly, hypertelorism, low-set ears, round face, acrochordons (skin tags) in front of eyes	Cat-like cry, hyperactivity, aggression, repetitive movements	Congenital heart disease, feeding difficulties	Speech impairment
Di George (velocardiofacial) syndrome	Deletion of the 22q11.2 region of chromosome 22	Borderline to mild	Hypertelorism, cleft palate	Speech and language deficits, inattention	Congenital heart disease, hypoparathyroidism, hearing impairment, recurrent infections, rheumatoid arthritis, renal abnormalities, feeding difficulties	Schizophrenia, early-onset Parkinson's disease, anxiety, depression, bipolar disorder
Down syndrome	Trisomy of chromosome 21	Mild to moderate	Brachycephaly, large tongue, epicanthal folds persisting beyond infancy, Brushfield spots, single palmar crease, small neck	Inattention, cheerful, sociable, reduced rates of maladaptive behaviours compared to peers with ID (23)	Congenital heart disease, hypothyroidism, gastro-oesophageal reflux, coeliac disease, leukaemia	Early-onset Alzheimer's-type dementia, obsessive-compulsive symptoms, autistic spectrum disorder
Fragile X syndrome	Trinucleotide (CGG) repeat mutation in FMR1 gene on X chromosome	Mild to moderate	Long face, large ears, prominent jaw and forehead, joint hyperextensibility, macroorchidism in males (post-puberty)	Shyness, poor eye contact, inattention, impulsivity, hyperactivity	Seizure disorder, infertility (female patients), strabismus	Autistic spectrum disorder, social anxiety, ADHD, OCD
Klinefelter syndrome	An additional X chromosome (47, XXY). Please note that other variants can exist (such as 48, XXXY and 49, XXXXY)	Normal intellectual functioning to mild	Tall stature, gynaecomastia, microorchidism	Inattention,	Infertility, gynaecomastia, osteoporosis, thromboembolism	Dyslexia
Lesch-Nyhan syndrome	Hypoxanthine-guanine phyosphoribosyltransferase (HPRT) mutation on X chromosome	Moderate	Coarse facial features, short thumbs and great toes	Self-mutilation, involuntary muscle movements, hypotonia	Gout, renal and bladder calculi	Severe self-injury
Neurofibromatosis type 1	Mutations in the neurofibromin 1 (NF-1) gene	Normal intellectual functioning to mild	Café au lait spots, neurofibromas, macrocephaly	No characteristic behavioural phenotype	Increased cancer risk, scoliosis, hypertension, optic gliomas, epilepsy	ADHD, social anxiety, depression (24)
Phenylketonuria	Mutations in the gene for phenylalanine hydroxylase, which converts phenylalanine to tyrosine	Mild to severe	Often normal appearance, may have lighter skin than other family members	Motor dysfunction, impaired executive function, challenging behaviour	Epilepsy, eczema, mouse-like odour	Autism spectrum disorder, ADHD, Parkinson's disease (25)
Prader-Willi syndrome	Lack of paternal contribution to a portion of chromosome 15	Mild-moderate	Narrow forehead, almond-shaped eyes, triangular (downturned) mouth, short stature, light skin and hair	Hyperphagia, skin picking, aggression, stubbornness	Obesity, hypogonadism, infertility, scoliosis, type 2 diabetes	Compulsive behaviour, sleep disturbances
Rett syndrome	Mutation in MECP2 gene on X chromosome	Severe	Often normal appearance, may have microcephaly	Repetitive stereotypic hand movements (e.g. hand wringing),	Encephalopathy, seizures, scoliosis, cardiac abnormalities	Autistic-like features, sleep disturbances

Table 2.3 Continued

Genetic syndrome	Underlying genetic pathology	Degree of ID	Morphological features	Behavioural phenotype	Physical associations	Psychiatric associations
Smith-Magenis syndrome	Deletion of p11.2 region of chromosome 17	Moderate	Square face, deep-set eyes, full cheeks, large jaw	Self-hugging, aggression, hyperactivity, inattention, impulsivity, self-injury	Dental abnormalities, scoliosis, visual and hearing impairment	Self-injury, sleep disturbances
Turners syndrome	Absence or partial absence of one X chromosome (45, X)	Normal intellectual functioning to mild	Webbed neck, broad chest, widely-spaced nipples, low-set ears, lymphoedema of peripheries	No characteristic behavioural phenotype	Infertility, congenital heart disease, osteoporosis, hypothyroidism	ADHD (26)
Williams syndrome	Deletion of q11.23 region of chromosome 7	Mild to moderate	Broad forehead, short nose, full cheeks, wide mouth	Lack of social inhibition, hyperactivity, affectionate demeanour	Supravalvular aortic stenosis, strabismus, gastrointestinal and renal problems	ADHD, anxiety, phobias

ID. Obtaining input from individuals themselves will be critical to the assessment process but it is recommended that a person's adaptive behaviour should be assessed through involvement of multiple third-party respondents and multiple sources of information including background information such as schooling and development including evidence for deficits reported (8).

Many factors may impact on the assessment, and consequently result in false positive or false negative results. These may include underlying mental health problems, use of pharmacological agents or illicit substances etc. Individuals suffering from active symptoms of mental illness will neither be able to engage fully in the process nor be functioning at optimal level. There is also evidence that many pharmacological agents, including prescription and illicit substances can influence neuropsychological functioning, especially sedating psychotropic medication. It is generally accepted that medication will have greatest effect in the first few weeks of prescription and for two weeks after withdrawal, and this should be taken into account. Motor impairments such as seen in those with cerebral palsy may lead to significant impairments in adaptive functioning which may be accounted for by motor impairment rather than ID. Another confounding factor might be motor impairments such as cerebral palsy and presence or absence of any neurodevelopmental disorders. The impact of neuro development disorders and adaptive functioning has also been studied; there are significant differences in the pattern of deficits in those with neuro development disorders such as autism. Adaptive behaviour in people with autism shows disproportionate deficits in socialization and communication domains compared to comparison groups, and this pattern of adaptive behaviour, without impairments in other areas may result in false positive results if clinicians do not consider this. Adaptive functioning is generally lower in children with autism, relative to IQ matched comparison groups, and this discrepancy between intelligence and adaptive behaviour found in autism should be taken into consideration during the assessment of intellectual disability. It is well known that slowed processing speed and working memory deficits are a common characteristic of variety of neurodevelopmental disorders such as attention deficit hyperactivity disorder (ADHD), and this may in turn impact on the overall intellectual functioning and adaptive functioning of the individual (2).

Intellectual functioning and adaptive behaviour are not highly correlated (7), and clinicians should take into account that people with IQ below 70 may function adequately and that there are many people with IQs above 70 with additional neurodevelopmental disorders such as autism, who have significant functional impairments (9). Understanding an individual's distribution of adaptive behaviour scores is a key task in order to recognize individual needs of patients, and should be used to inform decisions about care.

Working with different groups of patients

Patients may present to an assessment in a range of different ways, and one needs to endeavour to nevertheless make the assessment process valuable and meaningful for them. In all of the examples outlined below, it is essential to explore the reasons why the patient may be presenting as they are, as through better understanding of this, we can work on addressing such issues with a more informed approach. Carer involvement can be instrumental in this regard, as they may not only understand the basis for the patient's presentation, but also of successful techniques previously used to understand and remedy such situations.

Significant agitation

In patients presenting with significant agitation, distraction techniques such as engaging the patient in a discussion about their favourite subject, can be helpful in reducing their distress and fostering a supportive atmosphere. You need to be mindful of not encroaching on their personal space, and respecting the fact that an assessment can in itself be a stressful experience for patients, adding to their agitation (indeed, in some cases, the assessment may be the underlying source of the patient's agitation).

Hyperactivity

Though not overtly agitated, some patients may present as hyperactive, often manifesting in ways such as pacing around the room or fidgeting while seated. Your approach would be similar to that for an extremely agitated patient, as well as discussing with carers.

Aggression

As with any patient, it is crucial to consider any risks prior to the assessment, and in patients who have historical risks, have strategies in place to minimize their impact. Again, respecting the patient's personal space can be an effective strategy, as well as reducing the degree of direct eye contact with the patient, so as not to appear confrontational. For home visits, ensure that you are accompanied by a colleague, as well as informing your workplace base beforehand of where you are going and how you can be contacted. The room within which you are conducting the assessment should be carefully determined, with clear potential exit pathways identified. It is imperative to avoid a situation where the patient is situated between yourself and the exit pathways for the room. If the assessment is conducted in a clinical environment, panic alarms are often available—it is important to check that the alarm is functioning correctly prior to the assessment commencing.

Autistic spectrum disorder

Despite conventional wisdom, there is no one approach for interviewing patients with Autistic Spectrum Disorder (ASD). As Lorna Wing described, some patients may be 'passive' or 'aloof', and any attempted physical contact, such as shaking their hand for example, might actually be detrimental to rapport. Other ASD patients may be 'active and odd', and can conversely be overly familiar and invade your own personal space (10). Additionally, patients with ASD may be hypersensitive to sensory stimuli, so it is key to avoid the room being too brightly lit or having extraneous sounds present (indeed, some patients will be markedly distressed by something as seemingly innocuous as the ticking of a wall clock). Though starting an assessment by discussing a topic of the patient's interest is a generally useful approach in fostering rapport, this is particularly the case for those with ASD, who often have intensely observed special interests. An additional consideration would be to assess the patient at their own home, as ASD patients are likely to be distressed by an unfamiliar environment.

Sensory impairment

Please refer to Chapter 24 on Sensory Impairment for further information pertaining to working with patients with ID and sensory impairments. When patients have sensory impairments, adaptations should be made to the assessment approach so that the means of communication with the patient is that which they are most comfortable and proficient with. Liaise with any carers beforehand regarding how they communicate with the patient, as well as ascertaining the severity of any sensory impairment and whether any other sensory modalities are concomitantly affected. The involvement of a speech and language therapist from an early stage can help greatly, both in facilitating communication during the assessment as well as potentially working with the patient in the longer term. This may include developing a communication passport to further inform others how best to communicate with the patient. Finally, it is also important to appreciate that sensory impairments are often missed in PWID (particularly severe ID), and can be misconstrued by others; for example, someone with hearing impairment may be seen as not wanting to listen or having poor attention (11). Undiagnosed sensory impairment can have a major impact on the patient's ability to compensate for their ID and further reduce their potential; if such impairments are suspected, promptly refer the patient for further specialist sensory assessment to determine the presence and extent of any impairment. Ophthalmology and audiology services have modified assessment procedures for PWID, but the presence of a carer and a community intellectual disability nurse can also help support the patient as well as offering further relevant information to the assessor.

Profound and multiple disabilities

For patients with profound and/or multiple disabilities, liaising with well-established carers is again of utmost importance, as they will be best positioned to advise on how the patient communicates. This may be in the form of subtle or seemingly non-specific gestures that might otherwise go by unnoticed by even the more keen-eyed clinician. Despite the best possible efforts, you will likely be more dependent on carer testimony in this group of patients, though the importance of being informed and utilizing your observational skills cannot be overstated.

Mental state examination

Introduction

Obtaining a good history from the patient as well as from the carers and carrying out a good mental state examination is vital to the process of diagnostic formulation of mental health problems in any PWID. This process can however pose certain challenges in PWID. This could be attributable to communication issues, sensory impairments, the environment in which the assessment is carried out, and the presence of other comorbidities such as ASD.

Equal importance must be levied on the setting in which the mental state examination is carried out. It is often found that when the person is assessed in their own natural environment such as their family homes, residential home or the day centre, they tend to be more relaxed compared to having to attend a hospital setting to see the psychiatrist. This is more so for people with ASD, who often find new settings anxiety-provoking.

Just as in the general adult population, every effort needs to be made to see the PWID separately. If communication issues are identified, enlisting the help of a speech and language therapist can assist in improving the quality of the mental state examination and the psychiatric interview. As highlighted before, the use of special communication apps, picture exchange cards, Makaton, British Sign Language, and often simple use of pictures can assist with the interview process.

Components of Mental State Examination (MSE)

Appearance and behaviour

Clothing, gait, rapport, use of aids such as hearing aids, walking aids, or helmet for epilepsy need to be commented upon. Special comments need to be made regarding any dysmorphic features associated with genetic disorders. Any abnormal movements and stereotypes should also be noted. In people with ASD, eye contact and general behaviour should be commented upon. Some patients, particularly those with severe ID may be wheelchair-bound. Some may also bring objects which are of value to them in their lives, such

as a favourite toy, pieces of jewellery etc. People with ASD may be very particular about their personal belongings, or wear the same piece of clothing repeatedly irrespective of the weather.

Speech

Make sure to comment on the rate, tone, volume, and fluency in patients who are verbal. It is important to note for echolalia and the tone, especially in people with ASD. It is again useful to have an understanding of the patient's normal speech patterns, to identify any differences in the quality of the speech. For example, a patient with ASD may have a history of always speaking in whisper, and this could easily be mistaken for low volume secondary to depression. For people with other speech problems such as dysarthria or dysphasia, having carers who can understand and help with understanding the speech can also lead to vital clues in the mental state examination. Sometimes the clinician may not be able to comprehend the patient's speech and it is acceptable to acknowledge that and clarify what the patient is trying to say with the help of carers.

Mood

People with mild to moderate ID can often express their mood states. Nevertheless, some patients may struggle, and as such, using simple pictures of facial expressions (e.g. smiley face, sad face) can be of value. Across the ranges of ID, it is vital to obtain corroborative history from carers regarding the shift from the normative state, engagement in activities, interaction with family, carers, and any other behavioural changes. An example of this could be refusal to attend the day centre. Changes in biological functions such as sleep and appetite can be discerned through history and/or observation.

Affect

As in the general adult population, observe for range, congruency, and fluctuation in affect.

It may be difficult to accurately describe affect in people who have facial dysmorphic features due to an underlying genetic condition. For example, people with Angelman syndrome can always present as appearing happy, and this therefore will influence the description of affect. People with cerebral palsy can have a poor facial muscle tone and this again can make description of affect unreliable.

Thought

Every aspect of thought, such as content and form needs to be commented upon in the mental state examination in people who are verbal. It may sometimes be difficult to tease out the difference between overvalued ideas and delusions, and having a good background history of the patient can prove valuable in making this distinction. In addition, delusions usually lead to clear actions, which may not be observed with overvalued ideas. It is difficult to substantiate a diagnosis of schizophrenia in a patient with moderate/severe/profound ID, as some of the critical first rank symptoms cannot be elicited. In people with mild to moderate degrees of ID, delusions of reference, persecution, and grandeur can be identified; however, it may be difficult to identify delusions of guilt. Other thought abnormalities, such as flight of ideas can be identified in people with reasonably good verbal skills. People with ID can present with magical thinking and the level of their cognitive abilities need to be taken into consideration prior to establishing it to be pathological.

It is absolutely crucial to check explicitly for thoughts of self-harm or harm to others. Obsessive-compulsive thoughts can occur in ASD and obsessive-compulsive disorder.

However, the key difference is that the obsessions in obsessive-compulsive disorder have an egodystonic nature, in contrast to the egosyntonic quality in ASD, where the affected individual often gains pleasure from routines being followed in a certain manner.

Perception

Depending on verbal ability, hallucinatory experiences such as auditory hallucinations can be identified. Patients can describe 'hearing voices', however it may be difficult to identify whether they are in the second or third person. It is crucial that the patient understands the questions that are being asked to explore this symptomatology reliably. Clinicians need to refrain from asking leading questions as patients with ID are suggestible. Other hallucinatory experiences, such as in the visual, tactile, olfactory, and gustatory modalities need to be specifically enquired about. In residential homes or inpatient settings when there is a patient who has a history of hearing voices, other patients can often mimic this response and can themselves complain of hearing voices. It is important to bear in mind that people with ASD and ID often tend to talk to themselves and this can be misinterpreted as 'hearing voices'. Establishing the chronology of these behaviours often help to distinguish between psychosis and other aetiologies, including normative behaviour for that person.

Insight

It is important to identify the patient's awareness of their mental health problems and acceptance of the need for the treatment. This may be difficult in people with moderate to severe degrees of ID. Additionally, many patients with ID are recipients of passive care and hence may accept medications without questioning, but this does not constitute informed consent. Many ID psychiatry services have information leaflets available that are designed for different developmental levels, and can help to enhance insight.

Cognition

Keeping in mind the developmental age of the person, other cognitive functions such as attention, concentration and memory can be assessed. It is important for the clinician to have a clear idea about the approximate developmental age of the patient and presence or absence of other comorbidities. Assessing for memory problems is considered separately under dementia in Chapter 9.

Diagnostic formulation

In the clinical practice of ID psychiatry, arriving at a diagnosis involves primarily obtaining history from the patient (depending on their verbal ability), obtaining corroborative history from carers and family, and observation of the patient.

For non-verbal patients, often the use of simple pictorial aids can help in assessing mental state. For example, for assessing mood, drawing happy or sad smiley faces, and asking the patient to point out how they feel may be helpful. Observing the patient may clarify any issues related to behaviours, including those deemed to be challenging. Certain examples of descriptions of behaviours by family or carers are 'challenging behaviour', 'he is trying to be difficult',