

Intellectual Disability (Mental Retardation)

Sarah E. Shea, MD,
FRCPC*

Author Disclosure

Dr Shea has disclosed no financial relationships relevant to this article. This commentary does not contain a discussion of an unapproved/investigative use of a commercial product/device.

Educational Gap

The approach to genetic testing in the evaluation of intellectual disability is changing with the introduction of newer techniques, such as microarray testing.

Objectives After reading this article, readers should be able to:

1. Understand the terminology and definitions relevant to individuals who are intellectually disabled.
2. Be aware of the epidemiology, clinical presentations, and causes of intellectual disability (ID).
3. Know how to carry out an evaluation for the cause of a child's ID, realizing that in many cases a cause cannot be identified, and know how to assess the child's strengths and weaknesses.
4. Be aware of the latest trends in genetic testing in identifying the causes of ID.
5. Know the conditions that commonly coexist with ID and know proper medical follow-up.
6. Understand what is involved in the education of children and adolescents with ID, as well as reasonable expectations.
7. Be aware of ways to help an intellectually disabled youth transition to adulthood.

Definitions

Intellectual disability is a general term for what has previously been described as mental retardation. ID is a lifelong condition characterized by significant impairment of cognitive and adaptive development owing to abnormalities of brain structure or function. ID is not a single entity, but rather a general symptom of neurologic dysfunction. At this time, the terminology used with respect to intellectual disability (ID) (mental retardation [MR]) is shifting. The term *mental retardation* is still in use in many settings, including by some clinicians, and is found in legal and public policies that determine eligibility for support; however, the use of the term *intellectual disability* as a direct substitute for *mental retardation* is increasing. The American Association on Mental Retardation has been re-

named the American Association on Intellectual and Developmental Disabilities (AAIDD), while emphasizing that the definition of ID is exactly the same as that for MR. The chief reason for the change is that the term mental retardation is perceived as being pejorative; this semantic change does not reflect a reframing of the condition.

It is anticipated the next iteration of the *Diagnostic and Statistical Manual of Mental Disorders* (DSM) will use a different term in place of MR; within this article, the term used is ID.

There are two commonly used formal definitions for ID/MR. The definition for MR in the fourth edition of the DSM (DSM-IV) and the AAIDD definition for ID are worded slightly differently, but are essentially the same. Both base the definition on three coexisting features: (1) significantly subaverage intellectual functioning accompanied by (2)

Abbreviations

AAIDD:	American Association on Intellectual and Developmental Disabilities
ADHD:	attention-deficit/hyperactivity disorder
CT:	computed tomography
DSM:	<i>Diagnostic and Statistical Manual of Mental Disorders</i>
FAPE:	free appropriate public education
FAS:	fetal alcohol syndrome
ID:	intellectual disability
IQ:	intelligence quotient
MR:	mental retardation

*Editorial board member; Dalhousie University, Halifax, Nova Scotia.

deficits or impairments in adaptive skills with (3) onset before 18 years of age.

Degrees of MR (ID) are described within the DSM-IV. The correlating adjectives (mild, moderate, severe, profound) often are used in summarizing test results, and it is helpful to understand what they represent. Within the DSM, the ranges are given as intelligence quotient (IQ) numbers, which are based on the mean population IQ of 100, and 1 SD equals 15 points on commonly used assessments. A score of fewer than 70 points, ie, more than 2 SDs below the mean, represents ID. Mild ID is defined when the IQ falls 2 to 3 SDs below the mean (55 to 70). In moderate ID, the IQ is 3 to 4 SDs below the mean (40 to 55); in severe ID, the IQ is 4 to 5 SDs below the mean (25 to 40); and in profound ID the IQ is more than 5 SDs below the mean (below 25).

The AAIDD definition of ID does not reference these categories. The emphasis within that framework is on the level of support needed by an individual in addition to limitations to cognition and adaptive functioning. A tool for classification that can be applied as an alternative is the International Classification of Functioning, Disability, and Health developed by the World Health Organization, which includes body function and structure, activities, social participation, and environmental factors.

The descriptor borderline, as in borderline intelligence, or borderline ID/MR, is used sometimes to describe the situation where the IQ falls 1.5 to 2.0 SDs below the mean (70.0 to 77.5). Persons with that profile may have significant differences in function, especially in academics, but are not generally regarded as having ID.

By convention, for all levels of ID, reduced IQ must be accompanied by deficits in adaptive functions, which include conceptual, social, and practical skills. Conceptual skills include language, literacy, and numeracy achievement, as well as understanding of time and money. Social skills include social judgment, interpersonal skills, and social problem solving. Practical skills include personal care and other activities of daily living, occupational skills, ability to negotiate the world safely and access transportation, and so forth.

Clinicians vary with respect to application of the term ID (or MR) depending on the age of the individual. Some prefer to use the term developmental delay or global developmental delay for children younger than 5 years and reserve ID or MR for older children. For some, that usage appears to be a matter of convention, whereas for others it is a reflection of the need to have confidence in the developmental trajectory of a child before applying the latter terms. There has been a definition given to global developmental delay by the American Academy

of Neurology and the Practice Committee of the Children Neurology Society, which has further complicated the understanding of terminology. Within that definition, a child must have significant delay (2 SDs or more below the mean on age-appropriate, standardized, norm-referenced testing) in any two or more areas of development, which means the term would apply to many children who do not go on to meet criteria for ID. Finally, many health care practitioners and families continue to use developmental delay rather than ID or MR on the basis of its being perceived as a more positive and less stigmatizing term.

An individual with ID has delayed development, that is, the pattern and sequence follow a typical order but at a slower rate and with limitations with respect to the final level of achievement. This pattern is different from atypical development, in which children exhibit behaviors that fall outside of the normal, or expected, range of development. Children with autistic spectrum disorders develop atypically, and have qualitative differences in their ways of communicating and relating socially. They may or may not also show delayed development or ID.

Epidemiology

Estimates of the prevalence of ID vary from 1% to 3%, with the higher numbers likely representing cohorts defined only by IQ. Reports of actual measured prevalence vary with differences in the populations studied, case definitions, and study design. Most individuals with ID have IQs that fall in the mild range. ID is found more commonly in boys than in girls in a 1.4:1.0 ratio. Based on current population figures and using a 1% prevalence, there are currently more than 6 million American children and more than 600,000 Canadian children with ID.

Clinical Presentation

The age at identification typically reflects the degree of severity; that is, severely affected children typically are identified earlier because they fail to meet expected milestones in the first 2 years of life. Children with mild ID may not be identified until they have been in school for several years. Gross motor development frequently is within normal expectations in children who eventually are given a diagnosis of ID, whereas receptive language delay will virtually always be present. (In preschool children, receptive language is the best predictor of intelligence.) Some children with ID will be identified because of associated medical conditions, such as genetic syndromes.

Causes

When ID has been identified, it is important to attempt to identify the cause (Table 1). Although it is not a common occurrence, there are situations in which a condition will be identified that will be treatable, such as certain in-born errors of metabolism. Identification of cause also may lead to recognition of associated health risks (eg, immune deficiency associated with 22q11 deletion syndrome) or have genetic implications for the affected individual and his family (eg, fragile X syndrome). Identification of a cause can also be helpful in allowing a family to understand the child and to plan for the future, and can assist families in accessing support systems, some of which are organized around specific etiologic conditions.

In some cases, identification of the cause will be straightforward. In many children, however, no specific etiology will be identified. In those situations, the hunt for causation should be understood as an ongoing process because newer technologies may reveal answers that were not obtainable earlier. An excellent example of this evolution has been the impact of advanced genetic testing, such as microarray, on helping our understanding of the causes of ID.

Table 1. Causes of Intellectual Disability

Prenatal	Infection (eg, toxoplasmosis, rubella, cytomegalovirus, herpes simplex virus)
	Genetic abnormalities
	Trisomy 21
	Fragile X
	Other (point mutations; chromosome deletions, duplications, rearrangements, etc)
	Toxins and teratogens
	Alcohol
	Drugs
	Radiation
	Congenital hypothyroidism
Perinatal	Inborn errors of metabolism
	Hypoxia
	Complications of prematurity
	Intracranial hemorrhage
Postnatal	Perinatal CNS infection
	Acquired brain injury
	CNS hemorrhage
	CNS infection
	CNS malignancy
	Severe environmental deprivation
	Severe malnutrition
	Toxins (lead, mercury)

CNS, central nervous system.

The literature varies widely with respect to the reported success rate for identifying the cause of developmental delay or ID. Some studies report yields in the neighborhood of 10% and others as high as 80%. (1) (2) Studies that cite a high percentage of successful determination of cause generally ascribe etiology based on history, including intrapartum asphyxia and antenatal toxin exposure (alcohol or drugs). Cause often is ascribed to cerebral dysgenesis when neuroimaging is used. It can be argued that the former is making assumptions that may or may not be valid and that the latter is describing associated findings rather than identifying the etiology of the problem and the cause of the brain abnormalities.

Prenatal causes and genetic differences specifically are the most common causes of ID. Fragile X syndrome is the most common inherited disorder known to cause ID. The syndrome occurs in approximately 1 in 3600 boys and 1 in 4000 to 6000 girls, and in some series accounts for 1% to 2% of the diagnoses in boys with ID. Males with fragile X syndrome typically have moderate ID, but the cognitive outcome varies from learning disability to severe ID with or without autism. Girls also can have symptomatic fragile X syndrome (ie, one X has the full mutation), but their cognitive and physical phenotypes typically are milder. Approximately 50% of girls with fragile X syndrome will have borderline intellectual function or mild ID.

Boys with fragile X syndrome often have physical features of enlarged ears, a long face with a prominent chin, and large testicles (with puberty). They also may have connective tissue problems, such as mitral valve prolapse, flat feet, and hyperflexible joints, along with ear infections and skeletal problems. Behavioral characteristics in boys with fragile X can include ADHD, a rapid and “mumbling” speech pattern, hand biting or flapping, and behaviors associated with autism spectrum disorders, such as poor eye contact and intense responses to sensory stimuli. Girls with fragile X syndrome may or may not show physical differences, such as prominent ears. Girls may have learning difficulties (especially in math), attention problems, shyness or social anxiety, language dysfunction, including selective mutism, affective and schizophrenia spectrum disorders, and autism spectrum disorders.

A relatively common cause of ID is trisomy 21, or Down syndrome, which remains the most common chromosome abnormality among live-born infants. A recent study found that the prevalence of Down syndrome in 10 American states among children and adolescents (0–19 years old) was 10.3 per 10,000 in 2002. (3) Intellectual function varies in persons with Down syndrome; most have a mild to moderate degree of ID. Some persons with Down syndrome have severe ID and also

may have autism, which further affects their adaptive development.

The most common teratogen causing ID is alcohol. Those affected are referred to as having fetal alcohol spectrum disorder. The impact of fetal alcohol exposure varies and does not necessarily reflect the degree of exposure in a linear fashion; however, current recommendations advise against drinking any alcohol during pregnancy. The timing, frequency, and dose are factors that affect outcome, but there appear also to be significant individual factors, such as the mother's age and genetic/metabolic background, maternal nutrition, social risk factors, and exposure to other toxins. The cognitive impact of fetal alcohol exposure is best defined in the population deemed to have the full fetal alcohol syndrome (FAS) on the basis of coexisting differences in growth, physical characteristics, and neurodevelopmental profile. Typical facial characteristics of FAS include a flattened philtrum, a thin and flattened upper lip, and short palpebral fissures. It is estimated that FAS occurs in 0.5 to 2.0 per 1000 births in the United States. Most children with FAS have mild ID, although some have learning disabilities instead, and some will have moderate ID. Adaptive function in children with FAS is often further compromised by coexisting ADHD and oppositional defiant disorder and poor social insight.

Determining the Cause

The history and physical examination remain the most critical elements in finding the cause of a child's ID, followed by genetic testing. The history should include a careful family history focused on identifying other individuals, including second- and third-degree relatives, who have similar conditions, as well as a review of prenatal, perinatal, and neonatal events. The past medical history of neurologic and other acute and chronic conditions and a careful review of systems and recording of the behavioral pattern and developmental trajectory are important. Certain behavior patterns may be clues to a genetic disorder (eg, disturbed sleep, self-hugging, finger licking, and page flipping found in Smith-Magenis syndrome).

The physical examination should be comprehensive, because clues to cause may be found in any system. Examples of findings that may suggest syndromes associated with ID include microcephaly, growth abnormalities, unusual facies, skeletal abnormalities, skin markings (eg, hypo- or hyperpigmented areas, adenoma sebaceum), and palatal, cardiac, genitourinary anomalies. A careful neurologic examination may reveal abnormal head size or localizing findings. The physical examination should be supplemented by specific testing of vision and hearing, both as part of the search for cause and because of the

higher prevalence of comorbid sensory deficits. If fundoscopy is not possible, referral for ophthalmologic examination is recommended, because findings such as retinitis or optic nerve abnormalities may assist with diagnosis.

When abnormal findings occur, they may immediately suggest a known syndrome or disorder (eg, Down syndrome). If not, the practitioner can seek support for pattern recognition using tools such as the Online Mendelian Inheritance in Man (OMIM) Web site (4) or refer for consultation with a geneticist. Just as with the behavioral profile, the neurodevelopmental profile can provide its own clue when looked at in combination with the history and physical findings. For example, children with Williams syndrome often have a characteristic learning pattern, with strengths in verbal short-term memory and language and considerable weakness in visual spatial construction. When the history and physical examination do not suggest the cause of a child's ID, further investigation should be offered to the family, with the emphasis on genetic assessment.

Metabolic Testing, Electroencephalography

Where appropriate for age or if further indicated by the history or examination, screening for thyroid dysfunction and lead should be done. Routine metabolic screening is not likely to be helpful if there is nothing further in the history to suggest a possible metabolic disorder. This fact is increasingly true now that most places in Canada and the United States provide at least some newborn screening for metabolic diseases, such as phenylketonuria.

Items in the history that indicate the need to rule out metabolic disorders include developmental regression, hypoglycemia, unusual odor, liver or cardiac dysfunction, seizures, waxing and waning level of function, suggestive family history, parental consanguinity, ataxia, severe hypotonia, and severe to profound levels of impairment. Routine electroencephalography is not recommended, and should be reserved for situations in which there is a history of possible seizures.

Genetic Differences

A normal examination does not rule out an underlying genetic difference, because the physical examination is normal in at least 40% of children with chromosomal abnormalities and ID. The availability of array genomic hybridization technology has added complexity to the question of what approach to use for genetic assessment of the child with ID. One approach for children with ID and a nondiagnostic history and physical examination has been to obtain a karyotype and molecular analysis for

fragile X syndrome. For children with nondiagnostic histories and normal examinations whose results on the karyotype and fragile X molecular analysis are normal, additional genetic evaluation with microarray had not been universally recommended.

The approach to genetic testing is changing, however. The yield of microarray in children with ID has been reported to be 14% in one review, and roughly twice that of karyotyping. (5) The likelihood of finding chromosomal abnormalities with microarray has also been reported to increase with the number of clinical abnormalities (physical findings, seizures, abnormalities of tone, and so forth). Both the American College of Medical Geneticists (6) and The Canadian College of Medical Geneticists (7) recommended in 2010 and 2009, respectively, that array genomic hybridization should be the first-line laboratory investigation for children with unexplained ID and should be done instead of karyotype and fluorescence in situ hybridization testing. They emphasize that this testing should be undertaken only after appropriate counseling, and that karyotyping remains a better option if there are reasons to suspect a common aneuploidy (eg, trisomy 21, XXY).

They suggest also that microarray is not the best first-line assessment if one is suspecting certain specific syndromes (eg, Williams) for which a fluorescence in situ hybridization assessment would be more cost-effective. Microarray can reveal otherwise unrecognized deletions, duplications, and rearrangements; however, interpretation of the findings is not straightforward, because these changes may be found in typically developing individuals, including family members in some situations, and it is not always clear that the discovered microdeletion or duplication is causative. When a genetic abnormality is detected in a child with ID, testing of parents and sometimes other family members may be needed to aid in interpretation of the finding. This testing may reveal genetic abnormalities in other family members and also may reveal nonpaternity.

In some cases, the family history is particularly suggestive of an inherited genetic disorder. If there are two or more affected first-degree relatives with ID, multiple second- and third-degree relatives affected, relatives who are affected in a pattern that suggests an X-linked inheritance, a family history of known chromosomal abnormalities, or a history of multiple pregnancy losses, genetic evaluation is indicated. In those situations, evaluation by a geneticist and advanced testing should be considered.

Genetic tests of any kind should not be undertaken until there has been full discussion with the family and, if appropriate, the child about the potential implications of abnormal results, because findings may have medical, emotional, financial, and social implications. Abnormal

results in a child may have major implications for other family members' health. For example, diagnosis of fragile X syndrome in a boy may uncover the risk that pre-mutation carriers in the family could develop the fragile X-associated tremor/ataxia syndrome or fragile X-associated primary ovarian insufficiency. Nonpaternity also can be discovered through genetic testing.

Neuroimaging

There is disagreement about the value of neuroimaging in children with ID and otherwise normal examinations. MRI has been reported to be abnormal in as many as 13.9% of children in this situation. (1) The findings are not typically such that they prove causation, however, and may lead to more questions than answers. Findings on imaging also rarely inform management or affect outcome. The author's practice is to obtain an MRI in this situation only on children with more severe degrees of ID or if MRI may help confirm a cause suggested by history.

Some children will have specific findings that are suggestive of identifiable underlying brain pathology. These conditions include structural differences, such as abnormal hair whorls, midline defects, hypo- or hypertelorism, and abnormal head size, especially microcephaly. Clues can include functional differences, such as hemiplegia or other localizing findings on the neurologic examination. For this group, neuroimaging should be considered. MRI is the recommended modality, as it will pick up cerebral dysgenesis better than computed tomography (CT). If there is no urgency for the results, it is suggested that the MRI be scheduled after 18 months of age, because the later timing may reveal changes that may be hard to identify earlier owing to immature myelination. Use of MRI rather than CT also eliminates concern about radiation exposure. For cranial CT scans, the hypothesized lifetime risk of increased cancer-related mortality is cited as 0.07% if done in a 1-year-old. (8) MRI scanning in children with ID often requires significant sedation or anesthesia, and the risks and benefits always should be considered.

Medical Follow-Up

When the cause of a child's ID cannot be determined at the time of the initial investigations, it is important to establish follow-up. Periodic reexamination may reveal changes that lead to diagnosis, such as the evolution of café-au-lait spots in neurofibromatosis. Differences in pubertal development, as well as changes in sensory function or in growth, may lead to diagnosis; moreover, new diagnostic modalities, such as in the field of genetics, are to be expected.

Medical follow-up for those with diagnosed causes also is critical, because new treatments for previously untreatable disorders evolve with time (eg, bone marrow transplants for some storage disorders); moreover, children with ID are much more likely to have comorbid conditions.

Table 2 lists common comorbidities for children with ID and their estimated prevalence based on published studies. Management of the comorbidities is important to maximize both health and development. When there is coexisting ADHD, for example, all therapeutic modalities can and should be used, including medication. It is not true that persons with ID cannot respond to stimulants for treatment of ADHD; however, diagnosing ADHD requires understanding of the child's cognitive level.

Autism is much more common in persons with ID and should be identified to direct intervention strategies better. Most persons with ID will have some disturbance of behavior or a mental health issue at some point in their lives. It is very important to monitor and intervene for these. There is a risk that medications will become the automatic strategy when behavior is challenging. Medications definitely have a role to play in helping persons with ID, but they should not replace other approaches. Behavioral, educational, recreational, and psychopharmacologic interventions may play a role in successful treatment. Some individuals with mild ID can benefit from cognitive behavioral therapy, and support from mental health professionals skilled in assisting persons with ID should be obtained.

Also common are sleep disorders, ranging from circadian rhythm disruptions, to sleep disruptions secondary to gastroesophageal reflux or apnea, to nonorganic sleep problems, such as cosleeping and bedtime resistance.

Assessment

Inherent in the definition of ID is the need for determination of intelligence as well as measurement of adaptive function. It is critical that both realms be assessed appropriately, taking into account the environment, language, health, emotional state, and physical limitations of the

individual being evaluated. Tests used should be those that have been widely studied and validated on a population that is an appropriate match for the child being assessed. Most commonly used for individualized assessment are the Wechsler intelligence scales. Children ages 3 to 7 years and 3 months are tested with the Wechsler Preschool and Primary Scale of Intelligence, Third Edition (WPPSI-III). Between 6 and 16 years the Wechsler Intelligence Scales for Children, Fourth Edition (WISC-IV) is often used. Individuals 16 years and older can be tested with the Wechsler Adult Intelligence Scale, Fourth Edition (WAIS-IV). There are a variety of other tools that may be used to accommodate individual needs, including ones for children with limited language development. Adaptive function can be assessed by using standardized tools, such as the Vineland Adaptive Behavior Scales–Second Edition (VABS-II) and the Adaptive Behavior Assessment System–Second Edition (ABAS-II).

Caution is recommended in interpreting assessments because overdiagnosis of ID can have a negative impact on the child and family. The predictive value of intelligence testing increases with a child's age and is significantly stronger after the age of 4 years. Factors that may make intelligence test results less predictive of outcome include the coexistence of an autistic disorder, as well as testing shortly after coming out of a situation of significant environmental deprivation. Limitations imposed by sensory impairment or motor impairments must be factored into testing as well. Testing should be administered by individuals who have experience with assessment in individuals so affected.

Assessment should not be limited to determining a child's weaknesses and deficits. Appropriate testing will profile areas of strengths and interests also, and should assist with planning logical and effective programming to optimize outcome. The adaptive component of the assessment can be particularly helpful for this purpose. An interdisciplinary approach to assessment, planning, and intervention is important as well. Children and youth with ID benefit from the support of a wide range of professionals, including psychologists, occupational therapists, physiotherapists, speech-language pathologists, social workers, community and recreation therapists, audiologists, generalist and specialty physicians, and teachers. Collaboration and good communication among professionals and recognition of the key role that families play in the success of their children is vital.

Education

Federal law in the United States protects the rights of all children with disabilities, including those with ID. The

Table 2. Comorbid Conditions

Autism	20%–30%
Attention-deficit/hyperactivity disorder	9%–15%
Sensory impairment	Mild ID, 2%; Severe ID, 11%
Cerebral palsy	Mild ID, 6%–8%; Severe ID, 30%
Psychiatric/behavioral disturbance	80%

Individuals with Disabilities Education Improvement Act (Public Law 108-446) (IDEA), formerly known as the Education for All Handicapped Children Act (Public Law 94-142), requires all states receiving federal funding for education to provide a free appropriate public education (FAPE) for all individuals with disabilities from age 3 to 21 years that is “designed to meet each child’s unique needs and prepare them for employment and independent living.” In Canada, there is no federal law addressing this issue, and individual provinces have the understood obligation to provide education for students with special needs under Section 15 of the Charter of Rights. Services in Canada for children before school entry are provided variably from budgets that support health, community services, or education, and differ widely with respect to availability and funding support.

There has been a conscious shift away from the model of separate education for individuals with ID. The rationale for educational inclusion is part of a broader philosophy of community inclusion, founded in the belief that all members of the community have value and should be included. Most American and Canadian school systems use a policy of “least restrictive environment,” working to include children and youth with ID in mainstream settings when and where possible. The educational outcome of this inclusion is still open to debate, because meta-analyses are inconclusive and it is hard to control for the many factors that can affect the experience of the student in any school environment. (9)

There is evidence of improved functional outcome with inclusion of individuals who have mild ID within the mainstream classroom, and philosophically there can be no justification for isolating any population of children completely from their peers. The most reasonable approach is to ensure that educational plans and placements fit the needs of the individual child.

Some children with ID are not best served in a setting focused on traditional academic activities and will have increased opportunities to build their functional and recreational skills by having at least part of their school day in settings that are less broadly inclusive. Comorbid conditions, such as behavioral challenges or sensory impairments, also may dictate the need for specialized educational settings.

Inclusion alone does not appear to achieve adequate social integration, especially in the middle and high school years. Children with ID tend to become increasingly isolated socially and sometimes rejected by peers as they move into the later elementary school years. Structured approaches that help students with and without ID relate positively to one another can improve children’s sense of belonging. As children with ID age, they appear to be more likely to find their deepest friendships among others

with identified special needs. Educational and recreational settings that bring children and youth with special needs together can foster those relationships. If special educational settings, such as small specialized classrooms, are felt to be the best match for a child’s learning needs, inclusion still can be optimized through regular participation in activities with other groups within the school community.

Along with the move toward inclusive education has been a move away from grade retention as a strategy for educating children in general, including those with ID. The effect of grade retention on outcome in children with ID has not been well studied, but in studies of general populations of children, retention has been shown to impair self-esteem and to leave children out of sync with their peers with respect to size and pubertal stage, while failing to accelerate academic progress. In the author’s experience, grade retention can erode peer relationships for children with ID if they have been succeeding in an inclusive model, and should be considered only when individual circumstances make it desirable, such as when a good friend is in the grade below. High school is a time that can be managed in a more flexible manner, because grade placement has less impact on class membership and peer contacts owing to the selective and individualized nature of timetables.

Expectations

Academic achievement can, to some degree, be predicted by the level of ID. Table 3 indicates typical expectations for individuals with different levels of functioning. It must be understood, however, that individual variation and comorbidities may influence these expectations, as will opportunities for learning and experiences. It is important to recognize the influence of expectations on outcome and to avoid limiting individuals with ID, and physicians must avoid conveying unduly negative outlooks that may unintentionally do harm. Moreover, it is imperative to help each person discover and develop his or her strengths and interests, because these attributes can have a lasting positive impact on outcome and quality of life.

Communication Skills

Support for developing communication skills is critical for optimizing outcome for individuals with ID and should be an active component of their educational programming. Periodic reassessment is needed, looking at articulation, comprehension, adequacy and clarity of verbal expression, and pragmatic use of language. Some children with ID will need to be provided with alternate routes of communication. These methods may include sign language, picture

Table 3. General Expectations for Outcome According to Level of ID

Mild ID	Good self-help skills Some academic skills (early to late elementary level) May achieve independent living and employability, but most will need some support in these domains as adults
Moderate ID	Can perform basic self-help skills Possible limited academic skills (early reading and math) Will need supportive living and structured/supervised employment and recreation
Severe ID	Self-help and daily living skills typically require supervision and support Will not live independently May succeed in sheltered work setting
Profound ID	Limited communication Will require support for self-help and daily living skills Will remain dependent for all or most activities

communication symbol systems, such as the Picture Exchange Communication System, or electronic aids. The broad availability of tools such as electronic portable media player/personal digital assistant/ Wi-Fi mobile platforms, along with the many applications that can support communication, are making alternative routes increasingly available.

Functional and Leisure Skills

As indicated in the wording of the Individuals with Disabilities Education Improvement Act, the ultimate goal of the education of children with special needs is to prepare them for employment and independent living. Although many individuals with ID will not achieve full independence or typical employment, the preparation they receive throughout their childhood and adolescence will assist them in achieving their optimal quality of life. To that end, education programs must look broadly at the skill sets needed to maximize autonomy within the home and community. Factors include self-help and home skills, personal skills, health care skills, and activities such as getting around the community. It has been shown that quality of life for adults with ID correlates in part with leisure time skills, and these should be cultivated actively. For those individuals with ID who have the needed foundation, education should include skills for the workplace and an exploration of vocational or technical interests. An excellent resource for families and schools and health care providers is the Adolescent Autonomy Checklist developed by the Youth in Transition Project (1984–1987), University of Washington Division of Adolescent Medicine, and based on a model developed by the Children's Rehabilitation Center at the University of Virginia (available at <http://www.aacpp.com/pdf/parents/English/Teens/Adolescent-Autonomy-Checklist.pdf>). This checklist lays

out a variety of skills that all persons should acquire to achieve full autonomy and can be used to set goals and chart progress.

Social skills are another critical part of programming for individuals with ID and should include the basics of social communication, manners, grooming, and creating and maintaining friendships. Spiritual development is another core component of the human experience that should be available to everyone, including those with ID. Health care providers can assist institutions of faith in working with children and youth with ID by helping them to know how best to understand, educate, and communicate with the child. Identifying the role of spirituality in the life of the person with ID and his or her family can also help the care team in understanding their sources of meaning, coping, and connection to the community.

Health Maintenance

Children, youth, and adults with ID are at increased risk for poorer health outcomes. Obesity, poor physical fitness, and poor maintenance of oral health are concerns. For some individuals, these problems are exacerbated by associated medical conditions, such as Prader-Willi or Down syndrome; however, lifestyle issues and reduced access and use of health maintenance services are more important factors. Community recreational sports programs often are not designed to include children and youth with ID, and alternatives may not be available. Many children develop sedentary lifestyles, often relying on electronic forms of entertainment and other forms of passive recreation. Obesity associated with poor eating habits and reduced caloric expenditure often is a problem. Health care providers should be cautious in interpreting growth curves that have been designed for subgroups, such as people with Down syndrome, because

the ranges on the growth curves reflect the actual population rather than the ideal, and include those who are obese. BMI and measurement of waist circumference should supplement these curves in assessing for obesity.

Encouraging families of individuals with ID to take a family approach to healthy active living and to look for recreational opportunities that give meaningful exercise can help prevent chronic health issues. Many areas now have programs for children and youth with developmental disabilities, such as the Special Olympics program, and these programs can provide both an excellent source of physical activity and opportunities for social development. Consultation with recreation therapists can be extremely helpful in helping families connect with such programs and by helping community programs become inclusive.

The dental health of individuals with ID is a particular concern. In some cases, poor oral health is attributable to failure to attend to typical dental maintenance. In others, it may relate to underlying health conditions, difficulties cooperating with dental examinations, or limited financial resources. Some individuals with ID will not be reliable reporters of discomfort, and behavioral deterioration or failure to thrive can be presenting concerns of serious dental disease.

Puberty and Sexuality

Puberty is often a time of concern for parents of children with ID. Most individuals with ID will go through the changes of puberty at the typical time, although associated medical conditions may affect pubertal timing, such as precocious puberty in association with some types of central nervous system abnormality, such as hydrocephalus. Most individuals with ID negotiate puberty with relatively little difficulty. Menstrual hygiene often is a concern for parents. In general, teens who are able to manage their toilet hygiene independently will be successful in managing menstrual hygiene. Reminders and prompts can be built into the individual's home and school schedule. Menstrual suppression, most commonly done with sustained oral contraceptives, is an option. Use of oral contraceptives in a more typical fashion that includes monthly menstruation can be helpful to reduce the mood variability if there is significant premenstrual behavioral change or distress. Use of oral contraception can increase the predictability of menses and reduce the amount of flow, which also may assist with management. Decisions regarding menstrual manipulation should look at the risks and benefits, recognizing that substitute decision making in this and any other situation must always focus on the well-being of the individual for whom the decisions are being made.

Sterilization of males or females with ID is rarely acceptable. Sterilization is not a reasonable choice for

menstrual management or a substitute for preventing sexual victimization of an individual with ID. There may be situations when an individual with ID will wish to choose sterilization as his or her contraceptive option, however, and if in those situations it is felt that informed consent can be given, sterilization could be an appropriate choice.

Sexuality education is more challenging in the population with ID and often needs to be individualized to match the level of understanding. A good resource for this education is the Web site for the National Dissemination Center for Children with Disabilities (<http://nichcy.org/>). Parents may need help to understand that their children and youth with ID have typical sexual feelings. Masturbation is often a concern, and physicians should ask about it proactively. The core of management is teaching about privacy and use of redirection where needed. Social stories and "the bathing suit rule" can be helpful for teaching about both masturbation and appropriate touching. The bathing suit rule posits that people should not touch others nor allow others to touch them in the areas that are normally covered by a bathing suit.

Parents are frequently concerned about the risk of sexual coercion or abuse of their children and youth with ID. There is a limited benefit demonstrated at this time from programs focused on teaching adults with ID to negotiate potential sexual coercion, and appropriate supervision remains important.

Transition Points

Just as puberty can bring new concerns for families, other types of transition can be challenging for persons with ID and those who support them. There are recognized big transitions, such as initial school entry and shifts between schools, and eventually from school to whatever follows. Smaller transitions, however, can be equally problematic, such as the beginning of each school year and even vacations or other breaks in routine. Families report or demonstrate increased levels of stress at such times, as do persons with ID. A proactive approach to transition planning and support often can prevent problems such as behavioral decompensation. New team members benefit from advanced information about the children and youth they will assist as well as time for preparing materials, communication supports, and behavioral strategies. Children and teens with ID may benefit from introductions to new personnel and environments before the actual event, for example, going to the new school building during summer to meet new teachers and check out the environs.

An excellent resource to support both care providers and families around transitions can be found through

the CanChild Centre for Child Disability research Web site (<http://www.canchild.ca/en/childrenfamilies/abouttransitions.asp>). Stewart et al (10) have published best practice guidelines built around six themes:

1. Collaborative initiatives and policies are necessary supports for the transition to adult life.
2. Building capacity of people and communities will enhance the transition process.
3. The role of the "navigator" within communities facilitates capacity building.
4. Information and resources are available to all involved in the transition process.
5. Education is a critical component of any transition strategy.
6. Ongoing research and evaluation provides the evidence needed for success.

Moving to Adulthood

By definition, ID is a condition that affects an individual across the life span. Persons with ID and their families should be encouraged to look ahead periodically and to be sure that the incremental steps that are needed to take them to the end points for which they are aiming are in place. It is helpful for families and their supporting teams to use tools like those at the CanChild Web site and the Adolescent Autonomy Checklist (11) to lay out next steps. Vocational assessment often is an important step during the later high school years, and typically is accessed through the education system or community services. There are also issues of financial planning and determination of the need for guardianship or power of attorney arrangements that should be discussed before they are needed. As the youth with ID moves into adulthood, the question of where he or she will reside in the future also will need discussion and planning. Child health professionals can support families and persons with ID by directing them to resources in the community that can help with these issues early. They can also work to ensure that the adult with ID has a supportive and defined medical home by facilitating transition of medical care. Receiving physicians may have less direct experience in caring for persons with ID, and pediatricians can help by connecting them with resources, such as the Consensus Guidelines for Primary Health Care of Adults with Developmental Disabilities, (12) or condition-specific guidelines, such as the one available for adults with Down syndrome.

Advocacy

Health care providers should advocate for this especially vulnerable population. We are living in a society that still

discriminates on the basis of perceived intellectual value. That viewpoint is not acceptable. We all have an obligation to ensure that every person, including those with ID, receives an appropriate education and optimal health care, that the rights to give consent and assent are always considered, and that everyone has the opportunity for a full and satisfying life at every stage of development. Pediatricians can help at the individual, family, and community levels by identifying areas where additional support or opportunities are needed and by collaborating with others to assist in meeting those needs.

Summary

- Clinicians should consider replacing their use of the term *mental retardation* with *intellectual disability* (ID).
- A careful history and physical examination remain the most helpful tools for determining the cause of a child's ID.
- Genetic testing may reveal the etiology of ID in at least 14% of children with nondiagnostic histories and physical examinations.
- Individuals who have ID are at risk for poorer health outcomes.
- Transitions are points of particular vulnerability for persons who have ID and their families.

References

1. Shevell M, Ashwal S, Donley D, et al; Quality Standards Subcommittee of the American Academy of Neurology; Practice Committee of the Child Neurology Society. Practice parameter: evaluation of the child with global developmental delay: report of the Quality Standards Subcommittee of the American Academy of Neurology and The Practice Committee of the Child Neurology Society. *Neurology*. 2003;60(3):367–380
2. van Kamebeek CD, Scheper FY, Abeling NG, et al. Etiology of mental retardation in children referred to a tertiary care center: a prospective study. *Am J Ment Retard*. 2005;110(4):253–267
3. Shin M, Besser LM, Kucik JE, Lu C, Siffel C, Correa A; Congenital Anomaly Multistate Prevalence and Survival Collaborative. Prevalence of Down syndrome among children and adolescents in 10 regions of the United States. *Pediatrics*. 2009;124(6):1565–1571
4. Online Mendelian Inheritance in Man (OMIM) Web site. Available at: <http://www.ncbi.nlm.nih.gov/sites/entrez?db=omim&TabCmd=Limits>. Accessed November 15, 2011
5. Zahir F, Friedman JM. The impact of array genomic hybridization on mental retardation research: a review of current technologies and their clinical utility. *Clin Genet*. 2007;72(4):271–287
6. Manning M, Hudgins L; Professional Practice and Guidelines Committee. Array-based technology and recommendations for utilization in medical genetics practice for detection of chromosomal abnormalities. *Genet Med*. 2010;12(11):742–745

7. Canadian College of Medical Geneticists. Use of array genomic hybridization technology for constitutional genetic diagnosis in Canada. CCMG Position Statement (Clinical). Available at: <http://ccmg-cgmn.org/policy.html#position>. Accessed November 15, 2011
8. Brenner D, Elliston C, Hall E, Berdon W. Estimated risks of radiation-induced fatal cancer from pediatric CT. *AJR Am J Roentgenol*. 2001;176(2):289–296
9. Lindsay G. Educational psychology and the effectiveness of inclusive education/mainstreaming. *Br J Educ Psychol*. 2007;77 (pt 1):1–24
10. Stewart D, Freeman M, Law M, et al. “The best journey to adult life” for youth with disabilities: an evidence-based model and best practice guidelines for the transition to adulthood for youth with disabilities. CanChild Centre for Childhood Disability Research. 2009. Available at: <http://transitions.canchild.ca/en/OurResearch/bestpractices.asp>. Accessed November 15, 2011
11. Adolescent Autonomy Checklist. Available at: <http://www.aacpp.com/pdf/parents/English/Teens/Adolescent-Autonomy-Checklist.pdf>. Accessed November 15, 2011
12. Sullivan WF, Heng J, Cameron D, et al. Consensus guidelines for primary health care of adults with developmental disabilities. *Can Fam Physician*. 2006;52:1410–1418. Available at: <http://www.cfp.ca/cgi/content/short/52/11/1410>. Accessed November 15, 2011

PIR Quiz

This quiz is available online at <http://www.pedsinreview.aappublications.org>. NOTE: Since January 2012, learners can take *Pediatrics in Review* quizzes and claim credit online only. No paper answer form will be printed in the journal.

New Minimum Performance Level Requirements

Per the 2010 revision of the American Medical Association (AMA) Physician's Recognition Award (PRA) and credit system, a minimum performance level must be established on enduring material and journal-based continuing medical education activities that are certified for *AMA PRA Category 1 Credit™*. To successfully complete 2012 *Pediatrics in Review* articles for *AMA PRA Category 1 Credit™*, learners must demonstrate a minimum performance level of 60% or higher on this assessment, which measures achievement of the educational purpose and/or objectives of this activity.

Starting with 2012 *Pediatrics in Review*, *AMA PRA Category 1 Credit™* can be claimed only if 60% or more of the questions are answered correctly. If you score less than 60% on the assessment, you will be given additional opportunities to answer questions until an overall 60% or greater score is achieved.

1. Which of the following factors is most emphasized in the definition of intellectual disability (ID), as determined by the American Association on Intellectual and Developmental Disabilities?
 - A. Age at which ID is diagnosed.
 - B. Concomitant medical conditions.
 - C. Intelligence quotient (IQ).
 - D. Level of support required to perform daily functions.
 - E. Underlying cause of ID.
2. An 8-year-old girl in your practice is seen for a health maintenance visit. Her physical examination is normal. Her mother tells you that her prior testing has revealed an IQ of 60. Under the *Diagnostic and Statistical Manual of Mental Disorders* categorization, which of the following would be the most appropriate classification for her intelligence?
 - A. Mild ID.
 - B. Moderate ID.
 - C. Normal intelligence.
 - D. Severe ID.
 - E. Profound ID.
3. The mother of the 8-year-old girl discussed in question 2 asks you about what she can expect regarding her daughter's functioning over the next few years. Which of the following are you most likely to include in your counseling?
 - A. A magnetic resonance image of the brain can help predict how she will perform in school.
 - B. Delayed puberty can be expected.
 - C. Physical exercise will be difficult and should be discouraged.
 - D. She should be placed in a separate classroom from her peers throughout the school day.
 - E. She will likely develop good self-help skills.

4. Which of the following statements is true regarding children who have ID?
- A. Girls are more affected than boys.
 - B. Gross motor development is typically abnormal.
 - C. Mildly affected children are usually diagnosed in the first few years of life.
 - D. Most individuals with ID have IQs in the mild range.
 - E. Treatable causes of ID can be identified in most cases.
5. You are evaluating a 5-year-old boy who has been diagnosed with ID, with an IQ in the moderate range. He has not undergone any testing for a cause. A complete history, including a detailed family history, and complete physical examination are unremarkable. In most cases, which of the following tests should be performed first?
- A. Electroencephalography.
 - B. Genetic investigation (Karyotype/fragile X or microarray).
 - C. Head computed tomography.
 - D. Serum amino acid profile.
 - E. Urine organic acid screen.

Parent Resources From the AAP at HealthyChildren.org

The reader is likely to find material to share with parents that is relevant to this article by visiting these links:

- <http://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Mental-Retardation.aspx>
- <http://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Mental-Retardation-and-Pervasive-Developmental-Disorders.aspx>
- <http://www.healthychildren.org/English/health-issues/conditions/developmental-disabilities/Pages/Outlook-for-Children-with-Mental-Retardation.aspx>

Intellectual Disability (Mental Retardation)

Sarah E. Shea

Pediatrics in Review 2012;33;110

DOI: 10.1542/pir.33-3-110

Updated Information & Services

including high resolution figures, can be found at:
<http://pedsinreview.aappublications.org/content/33/3/110>

References

This article cites 8 articles, 3 of which you can access for free at:
<http://pedsinreview.aappublications.org/content/33/3/110#BIBL>

Subspecialty Collections

This article, along with others on similar topics, appears in the following collection(s):

Current Policy

http://classic.pedsinreview.aappublications.org/cgi/collection/current_policy

Developmental/Behavioral Pediatrics

http://classic.pedsinreview.aappublications.org/cgi/collection/development:behavioral_issues_sub

Cognition/Language/Learning Disorders

http://classic.pedsinreview.aappublications.org/cgi/collection/cognition:language:learning_disorders_sub

Children With Special Health Care Needs

http://classic.pedsinreview.aappublications.org/cgi/collection/disabilities_sub

Permissions & Licensing

Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at:
<http://classic.pedsinreview.aappublications.org/site/misc/Permissions.xhtml>

Reprints

Information about ordering reprints can be found online:
<http://classic.pedsinreview.aappublications.org/site/misc/reprints.xhtml>

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™





Pediatrics in Review

An Official Journal of the American Academy of Pediatrics

Intellectual Disability (Mental Retardation)

Sarah E. Shea

Pediatrics in Review 2012;33;110

DOI: 10.1542/pir.33-3-110

The online version of this article, along with updated information and services, is located on the World Wide Web at:

<http://pedsinreview.aappublications.org/content/33/3/110>

Pediatrics in Review is the official journal of the American Academy of Pediatrics. A monthly publication, it has been published continuously since 1979. Pediatrics in Review is owned, published, and trademarked by the American Academy of Pediatrics, 141 Northwest Point Boulevard, Elk Grove Village, Illinois, 60007. Copyright © 2012 by the American Academy of Pediatrics. All rights reserved. Print ISSN: 0191-9601.

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™

