Intellectual disability refers to a group of disorders that have in common deficits of adaptive and intellectual function and an age of onset before maturity is reached.

Definition

Contemporary conceptualizations of intellectual disability emphasize functioning and social interaction rather than test scores. The definitions of intellectual disability by the World Health Organization International Classification of Diseases, Individuals with Disabilities Education Act (IDEA), the American Psychiatric Association (Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition [DSM-5]) and the American Association on Intellectual and Developmental Disabilities (AAIDD) all include significant impairment in general intellectual function, social skills, and adaptive behavior. This focus on the conceptual, social, and practical enables the development of individual treatment plans designed to enhance functioning. Consistent across these definitions is onset of symptoms before age 18 yr or adulthood or during childhood, even if the diagnosis is made later in life.

Significant impairment in general intellectual function refers to performance on an individually administered test of intelligence that is approximately 2 SD below the mean. For a test that has a mean of 100 and SD of 15, IQ scores below 70 would meet these criteria. If the standard error of measurement is considered, the upper limits of subaverage intellectual function may extend to an IQ of 75. Using a score of 75 to delineate intellectual disability might double the number of children with intellectual disability, but the requirement for impairment of adaptive skills limits the false positives. Children with intellectual disability often show a variable pattern of strengths and weaknesses. Not all of their partial scores on IQ tests fall into the significantly subaverage range.

Significant impairment in adaptive behavior reflects the degree that the cognitive dysfunction impairs daily function. Adaptive behavior refers to the skills that are required for people to function in their everyday lives. The AAIDD and DSM-5 classifications of adaptive behavior addresses 3 broad sets of skills: conceptual, social, and practical. Conceptual skills include language, reading and writing, money concepts, and self-direction. Social skills include interpersonal skills, personal responsibility, self-esteem, gullibility, naiveté, and ability to follow rules, obey laws, and avoid victimization. Representative practical skills are performance of activities of daily living (dressing, feeding, toileting and bathing, mobility), instrumental activities of daily living (e.g., housework, managing money, taking medication, shopping, preparing meals, using the telephone), occupational skills, and the maintenance of a safe environment. For a deficit in adaptive behavior to be present, a significant delay in 1 of the 3 areas must be present. The rationale for requiring only 1 of the 3 areas is the empirically derived finding that people with intellectual disability can have varying patterns of ability and may not have deficits in all 3 areas.

The requirement for adaptive behavior deficits is the most controversial aspect of the diagnostic formulation. The controversy centers on 2 broad areas: whether impairments in adaptive behavior are necessary for the construct of
intellectual disability and what to measure. The adaptive behavior criterion may be irrelevant for many children; adaptive behavior is impaired in virtually all children who have IQ scores <50. The major utility of the adaptive behavior criterion is to confirm intellectual disability in children with IQ scores in the 65-75 range. It should be noted that deficits in adaptive behavior are often found in disorders such as autism spectrum disorders (see Chapter 30) and attention-deficit/hyperactivity disorder (ADHD) (see Chapter 33) in the presence of typical intellectual function.

The issues of measurement are important as well. The independence of the 3 domains of adaptive behavior has not been validated with research. The relationship between adaptive behavior and IQ performance is insufficiently explored. Most adults with mild intellectual disability do not have significant impairments in practical skills. It should be noted that adaptive behavior deficits must be distinguished from maladaptive behavior (e.g., aggression, inappropriate sexual contact).

Onset before age 18 yr or adulthood distinguishes dysfunctions that originate during the developmental period. The diagnosis of intellectual disability may be made after 18 yr or childhood, but the cognitive and adaptive dysfunction must have been manifested before age 18 or adulthood (e.g., during “childhood”).

The term mental retardation should be cast aside because it is stigmatizing, has been used to limit the achievements of the individual, and has not met its initial objective of providing assistance to people with the disorder. The term intellectual disability is increasingly used in its place, but has not been adopted universally. In the United States, some existing laws and their attendant entitlements still use the term mental retardation. In Europe, the term learning disability is often used to describe intellectual disability. Global developmental delay is a term often used to describe young children whose limitations have not yet resulted in a formal diagnosis of intellectual disability; it is often inappropriately used beyond the point when it is clear the child has intellectual disability, usually age 3 yr. Developmental delay is a classification that may be used by IDEA until age 9 yr.

Etiology

There appear to be 2 overlapping populations of children with intellectual disability: mild (IQ 50-70), which is more associated with environmental influences, and severe (IQ <50), which is more frequently linked to biologic and genetic causes. Mild intellectual disability is 4 times more likely to be found in the offspring of women who have not completed high school than in women who have graduated. This is presumably a consequence of both genetic (children can inherit an intellectual impairment) and socioeconomic (poverty, malnutrition) factors. The specific causes of mild intellectual disability are identifiable in <50% of affected individuals. The most common biologic causes of mild intellectual disability include genetic or chromosomal syndromes with multiple, major, or minor congenital anomalies (velocardiofacial syndrome, Williams syndrome, Noonan syndrome), intrauterine growth restriction, prematurity, perinatal insults, intrauterine exposure to drugs of abuse (including alcohol), and sex chromosomal abnormalities. Familial clustering is common.

In children with severe intellectual disability, a biologic cause (most commonly prenatal) can be identified in more than 75% of cases. Causes include chromosomal (e.g., Down syndrome Wolf-Hirschhorn syndrome, deletion 1p36 syndrome) and other genetic and epigenetic disorders (e.g., fragile X syndrome, Rett syndrome, Angelman and Prader-Willi syndromes), abnormalities of brain development (e.g., lissencephaly), and inborn errors of metabolism or neurodegenerative disorders (e.g., mucopolysaccharidoses) (Table 36-1). Nonsyndromic severe intellectual disability may be a result of inherited or de novo gene mutations, as well as microdeletions or...
microduplications not detected on standard chromosome analysis. More than 400 genes may be associated with nonsyndromic intellectual disability, with many detected by exomic sequencing. These de novo point mutations may also cause other phenotype features such as seizures or autism; the absence of these features suggests more pleotropic manifestations of genetic mutations. Consistent with the finding that disorders that alter early embryogenesis are the most common and severe, the earlier the problem occurs in development, the more severe its consequences tend to be.

Table 36-1
Identification of Cause in Children with Severe Intellectual Disability

<table>
<thead>
<tr>
<th>CAUSE</th>
<th>EXAMPLES</th>
<th>PERCENT OF TOTAL</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chromosomal disorder</td>
<td>Trisomies 21, 18, 13, Deletion 1p36, Klinefelter syndrome, Wolf Hirschhorn syndrome</td>
<td>~20</td>
</tr>
<tr>
<td>Genetic syndrome</td>
<td>Fragile X syndrome, Prader-Willi syndrome, Rett syndrome</td>
<td>~20</td>
</tr>
<tr>
<td>Nonsyndromic autosomal mutations</td>
<td>Variations in copy number, de novo mutations in SYNGAP1, GRIK2, TUSC3, oligosaccharyl transferase, and others</td>
<td>~10</td>
</tr>
<tr>
<td>Developmental brain abnormality</td>
<td>Hydrocephalus ± meningomyelocele, lissencephaly</td>
<td>~8</td>
</tr>
<tr>
<td>Inborn errors of metabolism or neurodegenerative disorder</td>
<td>PKU, Tay-Sachs, various storage diseases</td>
<td>~7</td>
</tr>
<tr>
<td>Congenital infections</td>
<td>HIV, toxoplasmosis, rubella, CMV, syphilis, herpes simplex</td>
<td>~3</td>
</tr>
<tr>
<td>Familial intellectual disability</td>
<td>Environment, syndromic, or genetic</td>
<td>~5</td>
</tr>
<tr>
<td>Perinatal causes</td>
<td>HIE, meningitis, IVH, PVL, fetal alcohol syndrome</td>
<td>4</td>
</tr>
<tr>
<td>Postnatal causes</td>
<td>Trauma (abuse), meningitis, hypothyroidism</td>
<td>~4</td>
</tr>
<tr>
<td>Unknown</td>
<td>Cerebral palsy</td>
<td>20</td>
</tr>
</tbody>
</table>

CMV, Cytomegalovirus; HIE, hypoxic ischemic encephalopathy; HIV, human immunodeficiency virus; IVH, intraventricular hemorrhage; PKU, phenylketonuria; PVL, periventricular leukomalacia.


**Epidemiology**

The prevalence of intellectual disability depends on the definition, the method of ascertainment, and the
population. According to statistics, 2.5% of the population should have intellectual disability, and 75% of these individuals should fall into the mild to moderate range. Rates vary across populations. Globally, the prevalence of intellectual disability has been estimated to be approximately 16.41/1,000 persons in low income countries, approximately 15.94/1,000 for middle-income countries, and approximately 9.21/1,000 in high-income countries. Overall, intellectual disability occurs more in boys than in girls: 2 : 1 in mild intellectual disability and 1.5 : 1 in severe intellectual disability. In part this may be a consequence of the many X-linked disorders associated with intellectual disability, the most prominent being fragile X syndrome (see Chapter 81.5).

In 2009-2010 in the United States, approximately 463,000 or 0.9% of school-age children received services for intellectual disability in federally supported school programs. For several reasons, fewer children than predicted are identified as having mild intellectual disability. Because it is more difficult to diagnose mild intellectual disability than the more severe forms, professionals might defer the diagnosis and give the benefit of the doubt to the child. Other reasons that contribute to the discrepancy are use of instruments that underidentify young children with mild intellectual disability (Chapter 30), some children being diagnosed as having autism spectrum disorders and their intellectual disability not addressed, and a disinclination to make the diagnosis in poor or minority students because of previous overdiagnosis.

Young children might show cognitive limitations without significant delays in adaptive behavior. As a result, new cases of mild intellectual disability continue to be diagnosed among children up to 9 yr of age. Children with intellectual disability also may be incorporated into another diagnosis (e.g., autism, cerebral palsy). Furthermore, it is possible that the number of children with mild intellectual disability is actually decreasing as a result of public health and education measures to prevent prematurity and provide early intervention and head start programs. In fact, the number of school children who receive services for intellectual disability has decreased since 1999, but if developmental delay is included, the numbers have not changed appreciably.

Unlike mild intellectual disability, where the prevalence may be decreasing, the occurrence of severe intellectual disability has not changed appreciably since the 1940s and is 0.3-0.5% of the population. Many of the causes of severe intellectual disability involve genetic or congenital brain malformations that can neither be anticipated nor treated at present. In addition, new populations with severe intellectual disability have offset the decreases in the prevalence of severe intellectual disability that have resulted from improved healthcare. Although prenatal diagnosis and subsequent pregnancy terminations have resulted in a decreased prevalence of Down syndrome (see Chapter 81), and newborn screening with early treatment has virtually eliminated intellectual disability caused by phenylketonuria and congenital hypothyroidism, an increased prevalence of maternal prenatal drug use (see Chapter 96.4) and improved survival of very-low birthweight premature infants has counterbalanced this effect.

Pathology and Pathogenesis

The limitations in our knowledge of the neuropathology of intellectual disability are exemplified by the fact that 10-20% of brains of persons with severe intellectual disability appear entirely normal by standard neuropathologic study. The majority of brains of these persons show only mild, nonspecific changes that correlate poorly with the degree of intellectual disability. These changes include microcephaly, gray matter heterotopias in the subcortical white matter, unusually regular columnar arrangement of the cortex, and neurons that are more tightly packed than
usual. Only a minority of the brain shows more specific changes in dendritic and synaptic organization, with dysgenesis of dendritic spines or cortical pyramidal neurons, or impaired growth of dendritic trees. The programming of the central nervous system (CNS) involves a process of induction; CNS maturation is defined in terms of genetic, molecular, autocrine, paracrine, and endocrine influences. Receptors, signaling molecules, and genes are critical to brain development. The maintenance of different neuronal phenotypes in the adult brain involves the same genetic transcripts that play a crucial role during fetal development, with activation of similar intracellular signal transduction mechanisms. Several syndromes that were thought to involve complex chromosomal abnormalities are, in fact, caused by single-gene mutations involving induction. Rubinstein-Taybi syndrome (see Chapter 81), a disorder marked clinically by broad thumbs and great toes, characteristic facies, and severe intellectual disability, results from a mutation in the gene encoding for the transcriptional coactivator CREB-binding protein (CBP), a factor important in the control of gene expression in early embryogenesis.

Clinical Manifestations

Early diagnosis of intellectual disability facilitates earlier intervention, identification of abilities, realistic goal setting, easing of parental anxiety, and greater acceptance of the child in the community. Most children with intellectual disability first come to the pediatrician's attention in infancy because of dysmorphisms, associated developmental disabilities, or failure to meet age-appropriate developmental milestones. There are no specific physical characteristics of intellectual disability, but dysmorphisms may be the earliest signs that bring children to the attention of the pediatrician. They might fall within a genetic syndrome such as Down syndrome or be isolated, as in microcephaly or failure to thrive. Associated developmental disabilities include seizure disorders, cerebral palsy, hypotonia, and autism; these conditions are seen more commonly in conjunction with intellectual disability than in the general population.

Most children with intellectual disability do not keep up with their peers and fail to meet age-expected norms. In early infancy, failure to meet age-appropriate expectations can include a lack of visual or auditory responsiveness, unusual muscle tone (hypo- or hypertonia) or posture, and feeding difficulties. Between 6 and 18 mo of age, gross motor delay (lack of sitting, crawling, walking) is the most common complaint. Language delay and behavior problems are common concerns after 18 mo (Table 36-2). Earlier identification of atypical development is likely to occur with more severe impairments; and intellectual disability is usually identifiable by age 3 yr.

Table 36-2
Common Presentations of Intellectual Disability By Age

<table>
<thead>
<tr>
<th>AGE</th>
<th>AREA OF CONCERN</th>
</tr>
</thead>
<tbody>
<tr>
<td>Newborn</td>
<td>Dystemorphic syndromes, (multiple congenital anomalies), microcephaly</td>
</tr>
<tr>
<td></td>
<td>Major organ system dysfunction (e.g., feeding and breathing)</td>
</tr>
<tr>
<td>Early infancy (2-4 mo)</td>
<td>Failure to interact with the environment</td>
</tr>
<tr>
<td></td>
<td>Concerns about vision and hearing impairments</td>
</tr>
<tr>
<td>Later infancy (6-18 mo)</td>
<td>Gross motor delay</td>
</tr>
<tr>
<td>Toddlers (2-3 yr)</td>
<td>Language delays or difficulties</td>
</tr>
</tbody>
</table>
For some children with mild intellectual disability the diagnosis remains uncertain during the early school years. It is only after the demands of the school setting increase over the years, changing from “learning to read” to “reading to learn,” that the child's limitations are clarified.

Adolescents with mild intellectual disability can present a diagnostic challenge. Typically they are up to date on current trends and are conversant as to who, what, and where. It isn't until the “why” and “how” questions are asked that their limitations become apparent. If allowed to interact at a superficial level, their mild intellectual disability might not be appreciated, even by professionals who may be their special education teachers or healthcare providers. Because of the stigma associated with intellectual disability, they may use euphemisms to avoid being thought of as “stupid” or “retarded” and refer to themselves as learning disabled, dyslexic, language disordered, or slow learners. Some people with intellectual disability emulate their social milieu to be accepted. They may be social chameleons and assume the morals of the group to which they are attached. Some would rather be thought “bad” than “incompetent.”

Laboratory Findings

The most commonly used medical diagnostic testing for children with intellectual disability include neuroimaging; metabolic, genetic, and chromosomal testing; microarray analysis; and electroencephalography. These tests should not be used as screening tools for all children with an intellectual disability. In some children, there is a reasonable yield for testing, whereas in others the yield of <1% does not support its use. Decisions on diagnostic testing should be based on the medical and family history, physical examination, testing by other disciplines, and the family's wishes. Table 36-3 summarizes clinical practice guidelines that have been published and the yields of testing to assist in evaluating the child with global developmental delay or intellectual disability. Microarray analysis has replaced karyotyping as the preferred approach for children with multiple anomalies or a positive family history. Microarray analysis has the ability to discern abnormalities that are below the resolution of karyotyping. For example, deletion 1p36 syndrome, the most common subtelomeric microdeletion syndrome (1:5,000 births), accounts for approximately 1% of children with developmental disabilities and is characterized by failure to thrive, microcephaly, deep-set eyes, midface hypoplasia, broad nasal bridge, heart deficits, and CNS anomalies. Noncompaction cardiomyopathy and seizures are also noted. The diagnosis is made by standard chromosomes in only approximately 20% and requires fluorescent in situ hybridization or microarray comparative genomic hybridization methods for remaining patients. Microarray analysis may identify variants of unknown significance or benign variants, and therefore should be used in conjunction with a genetic consultation. Karyotyping has a role for children whose array analysis is unrevealing and concern is present for inversions, balanced insertions, and reciprocal translocations. Fluorescent in situ hybridization and subtelomeric analysis have been largely replaced by microarray analysis but continue to be used for specific indications. If microarray analysis is not diagnostic whole
Exome sequencing increases the diagnostic yield in many children with nonsyndromic severe intellectual disability.

Table 36-3
Suggested Evaluation of the Child with Intellectual Disability/Global Developmental Delay

<table>
<thead>
<tr>
<th>TEST</th>
<th>COMMENT</th>
</tr>
</thead>
<tbody>
<tr>
<td>In-depth history</td>
<td>Includes pre-, peri-, and postnatal events (including seizures); developmental attainments; and 3-generation pedigree in family history</td>
</tr>
<tr>
<td>Physical examination</td>
<td>Particular attention to minor or subtle abnormalities; neurologic examination for focality and skull abnormalities</td>
</tr>
<tr>
<td></td>
<td>Behavioral phenotype</td>
</tr>
<tr>
<td>Vision and hearing evaluation</td>
<td>Essential to detect and treat; can mask as developmental delay</td>
</tr>
<tr>
<td>Gene microarray analysis</td>
<td>A 7.8% yield overall (10% in syndromic and 6.5% in nonsyndromic intellectual disability)</td>
</tr>
<tr>
<td></td>
<td>Better resolution than Karyotype. May identify up to twice as many abnormalities as karyotyping. Excellent in detecting de novo microdeletions or microduplications</td>
</tr>
<tr>
<td>Karyotype</td>
<td>Yield 4% in global developmental delay/intellectual disability</td>
</tr>
<tr>
<td></td>
<td>Best for inversions and balanced insertions, reciprocal translocations, and polyploidy</td>
</tr>
<tr>
<td>Fragile X screen</td>
<td>Combined yield 2%</td>
</tr>
<tr>
<td></td>
<td>Preselection on clinical grounds can increase yield to 7.6%</td>
</tr>
<tr>
<td>X-linked candidate intellectual disability genes</td>
<td>May explain up to 10% of intellectual disability</td>
</tr>
<tr>
<td></td>
<td>Yield may be as high as 42% if there is a definite family history and as high as 17% from a possibly linked kindred</td>
</tr>
<tr>
<td>Exomic gene sequencing</td>
<td>Detects inherited and de novo point mutations especially in nonsyndromic severe intellectual disability</td>
</tr>
<tr>
<td>Neuroimaging</td>
<td>MRI preferred. Positives increased by abnormalities of skull contour or microcephaly and macrocephaly, or focal neurologic examination. Overall has a higher yield</td>
</tr>
<tr>
<td></td>
<td>Identification of specific etiologies is rare. Most conditions that are found do not alter the treatment plan. Need to weigh risk of sedation against possible yield</td>
</tr>
<tr>
<td>Thyroid (T₄, TSH)</td>
<td>Near 0% in settings with universal newborn screening program</td>
</tr>
<tr>
<td>Serum lead</td>
<td>If there are identifiable risk factors for excessive environmental lead exposure</td>
</tr>
<tr>
<td>Metabolic testing</td>
<td>Yield 0.2-4.6% based on clinical indicators and tests performed</td>
</tr>
<tr>
<td></td>
<td>Urine organic acids, plasma amino acids, ammonia, lactate, and a capillary blood gas. Focused</td>
</tr>
</tbody>
</table>
Molecular genetic testing for fragile X syndrome is appropriate for a boy with moderate intellectual disability, unusual physical features, and/or a family history of intellectual disability, or for a girl with more subtle cognitive deficits associated with severe shyness and a relevant family history. For children with a strong history of X-linked intellectual disability, specific testing of genes or the entire chromosome may be revealing. MECP2 (methyl CpG binding protein 2 [Rett syndrome]) testing should be considered in girls with moderate to severe disability.

A child with a progressive neurologic disorder, developmental regression, or acute behavioral changes needs metabolic investigation (urinary organic acids, plasma amino acids, blood lactate, lysosomal enzymes in lymphocytes), although many of these disorders are detectable as part of newborn screening; a child with seizure-like episodes should have an electroencephalography performed. Children with micro- or macrocephaly or changes in head growth trajectory or asymmetric head shapes, as well as those with new or focal neurologic findings, including seizures, should have a neuroimaging procedure.

MRI scans identify a significant number of subtle markers of cerebral dysgenesis in children with intellectual disability. Formes frustes of amino acid and organic acid disorders are associated with intellectual disability in the absence of the more commonly associated manifestations of behavior change, lethargy, and coma.

Some children with more subtle physical or neurologic findings can also have determinable biologic causes of their intellectual disability (see Chapter 83 ([#!content/3-s2.0-B978145577566800083?scrollTo=%23c00083])). How intensively one investigates the cause of a child’s intellectual disability is based on a number of factors:

<table>
<thead>
<tr>
<th>MECP2 for Rett syndrome</th>
<th>1.5% of females with severe intellectual disability</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>0.5% of males</td>
</tr>
<tr>
<td>EEG</td>
<td>May be deferred in absence of history of seizures</td>
</tr>
<tr>
<td>Repeated history and physical examination</td>
<td>Can give time for maturation of physical and behavioral phenotype. New technology may be available for evaluation</td>
</tr>
</tbody>
</table>

EEG, Electroencephalogram; CGH, comparative genomic hybridization; MECP2, methyl CpG binding protein 2; T<sub>4</sub>, thyroxine; TSH, thyroid-stimulating hormone.

intellectual disability than in a child with a severe intellectual disability.

Is there a specific diagnostic path to follow? If there is a medical history or a family history, or if physical findings pointing to a specific disorder, a diagnosis is more likely to be made. In the absence of these indicators, it is difficult to choose specific tests to perform.

Are the parents planning on having additional children? If so, one would be more likely to intensively seek disorders for which prenatal diagnosis or a specific early treatment option is available.

What are the parents' wishes? Some parents have little interest in searching for the cause of the intellectual disability and focus exclusively on treatment. Others are so focused on obtaining a diagnosis that they have difficulty following through on interventions until a cause has been found. The entire spectrum of responses must be respected, and supportive guidance should be provided in the context of the parents' education.

Differential Diagnosis

One of the important roles of pediatricians is the early recognition and diagnosis of cognitive deficits. The developmental surveillance approach to early diagnosis of intellectual disability should be multifaceted. Parents' concerns and observations about their child's development should be listened to carefully, because their observations have been found to be as accurate as developmental screening tests. Medical, genetic, and environmental risk factors should be recognized. Infants at high risk (prematurity, maternal substance abuse, perinatal insult) should be registered in newborn follow-up programs in which they are evaluated periodically for developmental lags in the first 2 yr of life; they should be referred to early intervention programs as appropriate. Developmental milestones should be recorded routinely during healthcare maintenance visits. The American Academy of Pediatrics has formulated a schema for developmental surveillance and screening. Whether developmental surveillance is a more effective technique for identifying than recognizing failure to meet age-appropriate milestones has not been clearly established.

Before making the diagnosis of intellectual disability, other disorders that affect cognitive abilities and adaptive behavior should be considered. These include conditions that mimic intellectual disability and others that involve intellectual disability as an associated impairment. Sensory deficits (severe hearing and vision loss), communication disorders, and poorly controlled seizure disorders can mimic intellectual disability; certain progressive neurologic disorders can appear as intellectual disability before regression is appreciated. More than half of children with cerebral palsy (see Chapter 598) or autism spectrum disorders (see Chapter 30) also have intellectual disability as an associated deficit. Differentiation of isolated cerebral palsy from intellectual disability relies on motor skills being more affected than cognitive skills and on the presence of pathologic reflexes and tone changes. In autism spectrum disorders, language and social adaptive skills are more affected than nonverbal reasoning skills, whereas in intellectual disability there are usually more equivalent deficits in social, fine motor, adaptive, and cognitive skills.

Diagnostic Psychologic Testing

The formal diagnosis of intellectual disability requires the administration of individual tests of intelligence and adaptive functioning.
The Bayley Scales of Infant Development (BSID-III), the most commonly used infant intelligence scale, assesses language, visual problem-solving skills, behavior, fine motor skills, and gross motor skills in children between 1 mo and 42 mo of age. A Mental Developmental Index (MDI) and a Psychomotor Development Index (PDI, a measure of motor competence) score are derived from the results. This test permits the differentiation of infants with severe intellectual disability from typically developing infants, but it is less helpful in distinguishing between a typical child and one with mild intellectual disability.

The most commonly used psychologic tests for children older than 3 yr of age are the Wechsler Scales. The Wechsler Preschool and Primary Scale of Intelligence, 4th edition (WPPSI-IV) is used for children with mental ages of 2.5-7.6 yr. The Wechsler Intelligence Scale for Children, 4th edition (WISC-IV), is used for children who function above a 6 yr mental age. Both scales contain a number of subtests in the areas of verbal and performance skills. Although children with intellectual disability usually score below average on all subscale scores, they occasionally score in the average range in 1 or more performance areas.

The most commonly used test of adaptive behavior is the Vineland Adaptive Behavior Scale (VABS), which involves semi structured interviews with parents and/or caregivers and teachers that assess adaptive behavior in four domains: communication, daily living skills, socialization, and motor skills. Other tests of adaptive behavior include the Woodcock-Johnson Scales of Independent Behavior–Revised, the American Association on Intellectual and Developmental Disability Adaptive Behavior Scale (ABS-2nd edition), and the Adaptive Behavior Assessment System (ABAS-2nd edition). There is usually (but not always) a good correlation between scores on the intelligence and adaptive scales. Basic adaptive abilities (feeding, dressing, hygiene) are more responsive to remedial efforts than is the IQ score. Adaptive abilities are also more variable, which can relate to the underlying condition and to environmental expectations. Although persons with Prader-Willi syndrome (see Chapter 81) have stability of adaptive skills through adulthood, those with fragile X syndrome may have increasing deficits over time.

Complications

Children with intellectual disability have higher rates of vision, hearing, neurologic, orthopedic, and behavioral or emotional disorders than do typically developing children. These other problems are often detected later in children with intellectual disability. If untreated, the associated impairments can potentially adversely affect the individual's outcome more than the intellectual disability itself.

The most common associated deficits are motor impairments, behavioral and emotional disorders, medical complications, and seizures. The more severe the intellectual disability, the greater are the number and severity of associated impairments. Knowing the cause of the intellectual disability can help predict which associated impairments are most likely to occur. Fragile X syndrome and fetal alcohol syndrome (see Chapter 106.2) are associated with a high rate of behavioral disorders; Down syndrome has many medical complications (hypothyroidism, celiac disease, congenital heart disease, atlantoaxial subluxation). Associated impairments can require ongoing physical therapy, occupational therapy, speech-language therapy, adaptive equipment, glasses, hearing aids, and medication. Failure to identify and treat these impairments adequately can hinder successful habilitation and result in difficulties in the school, home, and/or neighborhood environment.

Prevention
Examples of primary programs to prevent intellectual disability include:

- Increasing the public's awareness of the adverse effects of alcohol and other drugs of abuse on the fetus
- Preventing teen pregnancy and promoting early prenatal care
- Preventing traumatic injury by encouraging the use of guards and railings to prevent falls and other avoidable injuries in the home; using appropriate seat restraints when driving and wearing a safety helmet when biking or skateboarding; teaching firearms safety
- Preventing poisonings by teaching parents about locking up medications and potential poisons
- Encouraging safe sexual practices to prevent the transmission of diseases
- Implementing immunization programs to reduce the risk of intellectual disability caused by encephalitis, meningitis, and congenital infection

Presymptomatic detection of certain disorders can result in treatment that prevents adverse consequences. State newborn screening by tandem mass spectrometry (now including >50 rare genetic disorders in most states), newborn hearing screening, and preschool lead poisoning prevention programs are examples. Thyroid screening in a child with Down syndrome is an example of presymptomatic testing in a disorder associated with intellectual disability.

Treatment

Although intellectual disability is not treatable, many associated impairments are amenable to intervention and therefore benefit from early identification. Most children with an intellectual disability do not have a behavioral or emotional disorder as an associated impairment, but challenging behaviors (aggression, self-injury, oppositional defiant behavior) and mental illness (mood and anxiety disorders) occur with greater frequency in this population than among children with typical intelligence. These behavioral and emotional disorders are the primary cause for out-of-home placements, reduced employment prospects, and decreased opportunities for social integration. Some behavioral and emotional disorders are difficult to diagnose in children with more severe intellectual disability because of the child’s limited abilities to understand, communicate, interpret, or generalize. Other disorders are masked by the intellectual disability. The detection of ADHD (see Chapter 33) in the presence of moderate to severe intellectual disability may be difficult, as may be discerning a thought disorder (psychosis) in someone with autism and intellectual disability.

Although mental illness is generally of biologic origin and responds to medication, behavioral disorders can result from a mismatch between the child’s abilities and the demands of the situation, organic problems, and/or family difficulties. They may represent attempts by the child to communicate, gain attention, or avoid frustration. In assessing the challenging behavior, one must also consider whether it is inappropriate for the child's mental age, rather than the chronological age. When intervention is needed, an environmental change, such as a more appropriate classroom setting, may improve certain behavior problems. Behavior management techniques are useful; psychopharmacologic agents may be appropriate in certain situations.

Medication is not useful in treating the core symptoms of intellectual disability; no agent has been found to improve intellectual function. Medication may be helpful in treating associated behavioral and psychiatric disorders.
Psychopharmacology is generally directed at specific symptom complexes including ADHD (stimulant medication), self-injurious behavior and aggression (neuroleptics), and anxiety obsessive-compulsive disorder, and depression (selective serotonin reuptake inhibitors). Before long-term therapy with any psychopharmacologic agent is initiated, a short trial should be conducted. Even if a medication proves successful, its use should be reevaluated at least yearly to assess the need for continued treatment.

Supportive Care and Management

Each child with intellectual disability needs a medical home with a pediatrician who is readily accessible to the family to answer questions, help coordinate care, and discuss concerns. Pediatricians can have effects on patients and their families that are still felt decades later. The role of the pediatrician includes involvement in prevention efforts, early diagnosis, identification of associated deficits, referral for appropriate diagnostic and therapeutic services, interdisciplinary management, provision of primary care, and advocacy for the child and family. The management strategies for children with an intellectual disability should be multimodal, with efforts directed at all aspects of the child’s life: health, education, social and recreational activities, behavior problems, and associated impairments. Support for parents and siblings should also be provided.

Primary Care

For children with an intellectual disability, primary care has a number of important components:

- Provision of the same primary care received by all other children of similar chronological age (see Chapter 5 (#!/content/3-s2.0-B9781455775668000053?scrollTo=%23c000005))

- Anticipatory guidance relevant to the child's level of function: feeding, toileting, school, accident prevention, sexuality education

- Assessment of issues that are relevant to that child's disorder: e.g., examination of the teeth in children who exhibit bruxism, thyroid function in children with Down syndrome, cardiac function in Williams syndrome (see Chapter 108 (#!/content/3-s2.0-B9781455775668001083?scrollTo=%23c00108))

The American Academy of Pediatrics has published a series of guidelines for children with specific genetic disorders associated with intellectual disability (Down syndrome, fragile X syndrome, and Williams syndrome). Goals should be considered and programs adjusted as needed during the primary care visit. Decisions should also be made about what additional information is required for future planning or to explain why the child is not meeting expectations. Other evaluations, such as formal psychologic or educational testing, may need to be scheduled.

Interdisciplinary Management

The pediatrician has the responsibility for consulting with other disciplines to make the diagnosis of intellectual disability and coordinate treatment services. Consultant services may include psychology, speech-language pathology, physical therapy, occupational therapy, audiology, nutrition, nursing, and/or social work, as well as medical specialties such as neurodevelopmental disabilities, neurology, genetics, psychiatry, developmental-behavioral pediatricians, and/or surgical specialties. Contact with early intervention and school personnel is equally important to help prepare the child’s Individual Family Service Plan/Individual Educational Plan. The family should be an integral part of the planning and direction of this process. Care should be family centered and culturally
Periodic Reevaluation

The child's abilities and the family's needs change over time. As the child grows, more information must be provided to the child and family, goals must be reassessed, and programming needs should be adjusted. A periodic review should include information about the child's health status as well as the child's functioning at home, at school, and in other community settings. Other information, such as formal psychologic or educational testing, may be helpful. Reevaluation should be undertaken at routine intervals (6-12 mo during early childhood), at any time the child is not meeting expectations, or when the child is moving from one service delivery system to another. This is especially true during the transition to adulthood, beginning at age 14 yr as mandated by the IDEA Amendments of 2004. This transitioning should include the transfer of care to the adult healthcare system by age 21 yr.

Educational Services

Education is the single most important discipline involved in the treatment of children with an intellectual disability. The educational program must be relevant to the child's needs and address the child's individual strengths and weaknesses. The child's developmental level, the child's requirements for support, and goals for independence provide a basis for establishing an Individualized Education Program for school-age children, as mandated by federal legislation.

Leisure and Recreational Activities

The child's social and recreational needs should be addressed. Although young children with intellectual disability are generally included in play activities with children who have typical development, adolescents with intellectual disability often do not have opportunities for appropriate social interactions. Participation in sports should be encouraged, even if the child is not competitive, because it offers many benefits, including weight management, development of physical coordination, maintenance of cardiovascular fitness, and improvement of self-image. Social activities are equally important, including dances, trips, dating, and other typical social and recreational events.

Family Counseling

Many families adapt well to having a child with intellectual disability, but some have emotional or social difficulties. The risks of parents' depression and child abuse and neglect are higher in this group of children than in the general population. Among the factors that have been associated with good family coping and parenting skills are stability of the marriage, good parental self-esteem, limited number of siblings, higher socioeconomic status, lower degree of disability or associated impairments, parents' appropriate expectations and acceptance of the diagnosis, supportive extended family members, and availability of community programs and respite care services. In families in which the emotional burden of having a child with intellectual disability is great, family counseling, parent support groups, respite care, and home health services should be an integral part of the treatment plan.

Prognosis

In children with severe intellectual disability, the prognosis is often evident by early childhood. Mild intellectual
disability might not always be a lifelong disorder. Children might meet criteria for intellectual disability at an early age, but later the disability can evolve into a more specific developmental disorder (communication disorder, autism, slow learner, or borderline normal intelligence). Others with a diagnosis of mild intellectual disability during their school years develop sufficient adaptive behavior skills so that they no longer fit the diagnosis as adolescents, or the effects of maturation and plasticity can result in children moving from one diagnostic category to another (from moderate to mild retardation). Some children who have a diagnosis of a specific learning disability or communication disorder might not maintain their rate of cognitive growth and fall into the range of intellectual disability over time. By adolescence, the diagnosis has generally stabilized.

The apparent higher prevalence of intellectual disability in low and middle income group countries is of concern given the limitations in available resources. While community-based rehabilitation is being implemented in more than 90 countries, the efficacy of such programs has not been established.

The long-term outcome of persons with intellectual disability depends on the underlying cause, the degree of cognitive and adaptive deficits, the presence of associated medical and developmental impairments, the capabilities of the families, and the school and community supports, services, and training provided to the child and family (Table 36-4). As adults, many persons with mild intellectual disability are capable of gaining economic and social independence with functional literacy. They might need periodic supervision, especially when under social or economic stress. Most live successfully in the community, either independently or in supervised settings. Life expectancy is not adversely affected by intellectual disability itself. □

Table 36-4
Severity of Intellectual Disability and Adult Age Functioning

<table>
<thead>
<tr>
<th>LEVEL</th>
<th>MENTAL AGE AS ADULT * (hl0000275)</th>
<th>ADULT ADAPTATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild</td>
<td>9-11 yr</td>
<td>Reads at 4th-5th grade level; simple multiplication and division; writes simple letter, lists; completes job application; basic independent job skills (arrive on time, stay at task, interact with coworkers); uses public transportation, might qualify for driver's license; keeps house, cooks using recipes</td>
</tr>
<tr>
<td>Moderate</td>
<td>6-8 yr</td>
<td>Sight-word reading; copies information, e.g., address from card to job application; matches written number to number of items; recognizes time on clock; communicates; some independence in self-care; housekeeping with supervision or cue cards; meal preparation, can follow picture recipe cards; job skills learned with much repetition; uses public transportation with some supervision</td>
</tr>
<tr>
<td>Severe</td>
<td>3-5 yr</td>
<td>Needs continuous support and supervision; might communicate wants and needs, sometimes with augmentative communication techniques</td>
</tr>
<tr>
<td>Profound</td>
<td>&lt;3 yr</td>
<td>Limitations of self-care, continence, communication, and mobility; might need complete custodial or nursing care</td>
</tr>
</tbody>
</table>


For persons with moderate intellectual disability, the goals of education are to enhance adaptive abilities and “survival” academic and vocational skills so they are better able to live in the adult world (see Table 36-4).
The concept of supported employment has been very beneficial to these individuals; the person is trained by a coach to do a specific job in the setting in which the person is to work. This bypasses the need for a sheltered workshop experience and has resulted in successful work adaptation in the community for many people with an intellectual disability. These persons generally live at home or in a supervised setting in the community.

As adults, people with severe to profound intellectual disability usually require extensive to pervasive supports (see Table 36-4). These individuals may have associated impairments, such as cerebral palsy, behavioral disorders, epilepsy, or sensory impairments, that further limit their adaptive functioning. They can perform simple tasks in supervised settings. Most people with this level of intellectual disability are able to live in the community with appropriate supports.

Bibliography is available at Expert Consult.

Bibliography


Intellectual disability involves problems with general mental abilities that affect functioning in two areas: intellectual functioning and adaptive functioning. Learn more at psychiatry.org. What is Intellectual Disability? Intellectual disability involves problems with general mental abilities that affect functioning in two areas: intellectual functioning (such as learning, problem solving, judgement). adaptive functioning (activities of daily life such as communication and independent living). Intellectual disability is a condition in which your brain doesn’t develop properly or function within the normal range. Learn about symptoms and causes. What You Should Know About Intellectual Disability. Medically reviewed by Karen Richardson Gill, MD, FAAP. Written by Shannon Johnson. Updated on August 14, 2019. Symptoms. Levels of intellectual disability. Causes. Diagnosis.