Intellectual disability  Alternative Name
MENTAL RETARDATION

If a kid has an intellectual disability, it means that he or she learns and develops more slowly than other kids. At one time, intellectual disabilities were called "mental retardation," but that term is not used as much anymore because it hurts people's feelings.

Someone who has an intellectual disability will have trouble learning and functioning in everyday life. This person could be 10 years old, but might not talk or write as well as a typical 10-year-old. He or she also is usually slower to learn other skills, like how to get dressed or how to act around other people.

But having an intellectual disability doesn't mean a person can't learn. Ask anyone who knows and loves a person with an intellectual disability! Some kids with autism, Down syndrome, or cerebral palsy may be described as having an intellectual disability, yet they often have a great capacity to learn and become quite capable kids.

Just like other health problems, an intellectual disability can be mild (smaller) or major (bigger). The bigger the disability, the more trouble someone will have learning and becoming an independent person.

Mental Retardation: What it is and What it is Not

Overview

A diagnosis of mental retardation carries with it certain unique treatment needs that must be understood and addressed. Unfortunately, most psychiatrists are ill-equipped to handle this situation, having received little or no formal training in this area. This article is written with the specific goal of giving psychiatrists a better understanding of the special needs of patients with mental retardation and strategies for improving their quality of life.

Mental retardation is a state of developmental deficit, beginning in childhood, that results in significant limitation of intellect or cognition and poor adaptation to the demands of everyday life. As noted by Esquirol, intellectual disability is not a disease in and of itself, but is the developmental consequence of some pathogenic process.[1]

The Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition, Text Revision (DSM-IV-TR) defines mental retardation as follows:[2]

- Significantly subaverage intellectual functioning - An intelligence quotient (IQ) of approximately 70 or below
- Concurrent deficits or impairments in adaptive functioning in at least 2 of the following areas: communication, self-care, home living, social/interpersonal skills, use of community resources, self-direction, functional academic skills, work, leisure, health, and safety
Onset before age 18 years
Although mental retardation is classified as an axis II disorder in *DSM-IV-TR*, it is not considered a mental illness as such, with its own unique signs and symptoms. It is a system of identifying groups of people who need social support and special educational services to carry out tasks of everyday living.

The causes of mental retardation can be grouped from most to least common as follows:

- Alterations in embryonic development, such as those caused by chromosomal abnormalities or fetal exposure to drugs or toxins
- Environmental deprivation and other mental disorders, such as autism[^3,^4]
- Problems of pregnancy and the perinatal period, such as fetal malnutrition, hypoxia, infection, trauma, or prematurity
- Hereditary abnormalities, such as inborn errors of metabolism or chromosomal aberrations
- Medical conditions of infancy or childhood, such as central nervous system (CNS) infection or trauma, or lead poisoning

Physical causes are evident in most cases of moderate-to-profound retardation. A disadvantaged environment is more likely in mild retardation.

Many of the classification systems for mental retardation have been based on the timing of the insult to the CNS. The successive classification systems developed by the American Association on Mental Retardation also followed the timing approach.

**Prenatal Causes - Genetic Disorders**

Prenatal genetic disorders are characterized by changes in the genetic material, which may or may not have been inherited from the parents. Prenatal genetic disorders are characterized by changes in the genetic material, which may or may not have been inherited from the parents. Recent advances in the science and understanding of genetics have allowed a higher diagnostic yield from such testing.[^5]

**Chromosomal aberrations**

*Down syndrome* is the best-known example of a prenatal genetic disorder. In 95% of cases, Down syndrome is caused by trisomy 21, in which the extra chromosome 21 in the egg or sperm cell results from the nondisjunction in the meiotic stage. When such a gamete becomes fertilized, the fetus will have an extra chromosome 21 in all cells, for a total of 47 chromosomes.

In cases of Down syndrome caused by translocation, there are 46 chromosomes, but chromosomal material from 47 chromosomes is present because an extra chromosome 21 is attached (translocated) to another chromosome, usually chromosome 14 (designated as t(14;21)). In approximately half of translocation cases, a parent (usually the mother) has a balanced translocation, ie, 45 chromosomes with t(14;21). If a child has translocation Down syndrome, the parents should be examined for the presence of a balanced translocation. This is important in genetic counseling because when the mother or father has a t(14;21) translocation, the chance of having a child with Down syndrome is 1 in 10 if in the mother or 1 in 20 if in the father.[^6,^7,^8]

In another variant, mosaicism, some cells have 47 chromosomes and others have 46 because of an error in one of the first cell divisions of the fertilized egg. The characteristic phenotype of Down syndrome is basically the same in trisomy 21 and in translocation. The main features are upward-slanted palpebral fissures, a low nasal bridge with epicanthal folds, a small mouth and ears, a single palmar crease (simian crease), a flat nasal bridge, short and wide palms, and a characteristic dermatoglyphic pattern.
A loss of part of a chromosome is called a deletion. The best-known example is cri-du-chat syndrome, which is characterized by a high-pitched voice and is caused by a deletion in chromosome 5p3. Note that most fetuses with chromosomal aberrations are not viable. Approximately 40-50% of spontaneously aborted fetuses have a chromosomal anomaly. Only 2 of 10 fetuses with Down syndrome are born alive.

A new method of using DNA probes and fluorescence in situ hybridization has brought new light to many of the malformation syndromes previously classified as being of unknown origin. The same submicroscopic deletions (microdeletions) of DNA have been reported in chromosome 15q11-12 in the Prader-Willi and Angelman syndromes, despite the fact that these syndromes have different phenotypes. Because of the mechanism of imprinting, the Prader-Willi syndrome results when the microdeletion is in the chromosome of paternal origin and the Angelman syndrome results when it is of maternal origin.[9]

Persons with the Prader-Willi syndrome have an excessive appetite and indiscriminate eating habits, leading to obesity. Because this syndrome has no clear pathognomonic features, it may remain undiagnosed, and such individuals might even be referred for psychiatric treatment because of an eating disorder. Obviously, psychological factors are not the primary cause here, but supportive psychotherapy might be helpful. The treatment is based on behavioral modification, the institution of strict environmental limits on food intake, and necessary educational and habilitative programming. In addition, some studies have demonstrated that treatment with growth hormone improves somatic and behavioral deficits in these patients.[10, 11, 12, 13]

Disorders with autosomal-dominant inheritance

Tuberous sclerosis is an example of the disorders in this group, which might be associated with mental retardation. It is caused by a mutation in a gene affecting the formation of the ectodermal layer of the embryo. Because the skin and the CNS develop from this layer, abnormalities are seen in both.

The skin lesions include angiofibromas in the form of macules on the cheeks (adenoma sebaceum), with a butterfly-like distribution, especially after puberty. Café au lait spots or nonpigmented ash leaf–shaped areas are also found. Mental retardation, epilepsy, and calcifications in the brain are seen, as are tumors. Epileptic seizures often begin as infantile spasms, which should alert the physician to look for other symptoms of this disorder.

If tuberous sclerosis is diagnosed, both parents should be examined carefully because the mutation is inherited in approximately 28% of cases. Because of the dominant inheritance, the risk of recurrence is 50% for each pregnancy. The expression of this gene mutation varies from small skin discolorations (which may indicate a carrier state) to multiple disabling conditions. It is a relatively rare disorder (prevalence of 1 in 30,000 to 1 in 50,000 live births), but it may be found in approximately 0.5% of persons with severe mental retardation.[14]

Disorders with autosomal-recessive inheritance

Most metabolic disorders belong to this category. They are caused by single mutated genes that disturb the metabolism by deficient enzyme activity. The risk of healthy carrier parents having an affected child is 25% for each pregnancy. The diagnosis is made by detection of abnormal metabolic products in the urine, blood, or tissues and/or by low or absent enzyme activity.

Phenylketonuria (PKU) is the best known and most common of the metabolic disorders, with a prevalence of approximately 1 in 10,000 live births. The enzymatic defect is diminished activity of phenylalanine hydroxylase, which leads to a high serum phenylalanine level, affecting, among other things, myelination of the CNS. It was described in 1934 by Folling in 10 children with mental retardation, hypertonia, and hyperreflexia, with a musty odor in urine and sweat. Seizures and tremors are common, as are eczema and psychotic manifestations.
The clinical symptoms can be prevented by use of a low-phenylalanine diet soon after birth. In most developed countries, all newborns are screened for PKU. Increasingly, a lifelong low-phenylalanine diet is recommended to prevent later deterioration in cognitive functions. Women with PKU who were successfully treated do not have clinical manifestations themselves but still have phenylalanine blood levels high enough to cause brain damage to a fetus if they become pregnant. To avoid this, they should start to follow the diet again before they become pregnant.

**X-linked mental retardation**

Fragile X syndrome is the most common inherited form of mental retardation and, after Down syndrome, the most common genetic form. It is X linked, with dominant inheritance, and the penetrance is lower in females. Because of a constriction at the location Xq27.3, it appears as if the chromosome is fragile and a part of it is breaking off.

Prepubertal boys with this syndrome look quite normal. They often are restless and hyperactive and have a short attention span. Their developmental milestones, especially speech development, are delayed. After puberty, the characteristic phenotypical features may appear. They include an oblong face, prominent ears and jaw, and macroorchidism. Most have moderate mental retardation, but retardation is more severe in others. Male carriers do not have mental retardation. Females with fragile X syndrome who have the full mutation and are symptomatic usually have learning disabilities or mild mental retardation. Behavioral symptoms have been described in these individuals, ie, hyperactivity and social withdrawal in approximately 50% and depression in approximately 25%.

**Maternal infections**

Viral infections in the mother can interfere with organogenesis, and the earlier in pregnancy they occur, the more severe their effect will be, as exemplified by congenital rubella. Rubella infection during the first month of pregnancy affects the organogenesis of 50% of embryos. Infection in the third month of pregnancy still disturbs the development of 15% of fetuses. Various systems are affected, and as a result, symptoms and impairments may vary and include mental retardation, microcephaly, hearing and vision impairment, congenital heart disease, and behavior problems. Fortunately, the incidence of congenital rubella has greatly decreased because of the availability of immunization for prospective mothers.

Congenital cytomegalovirus infection may result in microcephaly, sensorineural hearing loss, and psychomotor retardation. Antibodies against cytomegalovirus are found in approximately 80% of adults. Depending on the population, primary infections occur during 2-5% of pregnancies. Cytomegalovirus inclusion bodies are seen in urine specimens of newborns who were infected prenatally.

Congenital toxoplasmosis may result in significant problems in approximately 20% of infected infants (eg, hydrocephalus, microcephaly, psychomotor retardation, vision and hearing impairment) and in milder developmental problems later in life.

Congenital human immunodeficiency virus infection has been increasing in importance. In a German study of 41 children born to mothers who were positive for human immunodeficiency virus infection, neurological symptoms were described at age 1-7 years. Human immunodeficiency virus encephalopathy was characterized by microcephaly, progressive neurological deterioration, mental retardation, cerebellar symptoms, and behavioral changes. Prophylactic intravenous immunoglobulin therapy with and without zidovudine often was able to prevent regression. Improvement was seen with zidovudine treatment.

**Toxic substances**

The most important of the teratogenic substances is ethanol, which is the cause of fetal alcohol syndrome (FAS). The prevalence of this syndrome varies around the world, but its occurrence in
industrialized countries is estimated to be approximately 1 in 1000 newborns. When used heavily during pregnancy, alcohol causes abnormalities in 3 main categories: (1) dysmorphic features, which originate in the period of organogenesis; (2) prenatal and postnatal growth retardation, including microcephaly; and (3) CNS dysfunction, including mild-to-moderate mental retardation, delay in motor development, hyperactivity, and attention deficit. The severity of the symptoms is related to the amount of alcohol ingested.

**Toxemia of pregnancy and placental insufficiency**

Intrauterine growth retardation has many causes, the most important being maternal toxemia with its consequences, ending in insult to the CNS. Prematurity may be of maternal or fetal origin. When it is connected with fetal developmental deviations, the prognosis depends on the infant's general condition. Prematurity and especially intrauterine growth retardation predispose to many perinatal complications, which may result in insult to the CNS and developmental problems.

**Perinatal Causes**

This period refers to 1 week before birth to 4 weeks after birth.

**Infections**

During the neonatal period, the most important infection, from the point of view of its developmental sequelae, is herpes simplex type 2. The neonate is infected during the delivery and may develop encephalitis within 2 weeks. Early treatment with acyclovir may alleviate the otherwise poor outcome, ie, microcephaly, profound mental retardation, and neurological deficits. Neonatal bacterial infections might result in sepsis and meningitis, which, in turn, may cause hydrocephalus.

**Delivery problems**

During delivery, asphyxia is the most important factor causing an insult to the CNS. It leads to cell death, which might be demonstrated with neuroimaging techniques as leukomalacia. Premature infants and those with intrauterine growth retardation are at special risk for damage to the cortex or thalamus, which, in addition to affecting intelligence, causes various symptoms of cerebral palsy (CP) and seizure disorder, depending on the location of the pathological condition. Importantly, note that asphyxia alone does not cause mental retardation.

Neurologic symptoms during the neonatal period have a strong association with prenatal developmental deviations and later neurologic integrity and intellectual level. For these reasons, infants with perinatal problems need a thorough examination for dysmorphic features and close follow-up because multiple disabilities might become evident later in life.

**Other perinatal problems**

Retinopathy of prematurity (formerly referred to as retrolental fibroplasia) was seen frequently when the use of 100% oxygen in neonates was common, resulting in blindness. It is often associated with other CNS damage, mental retardation, and other developmental problems. Infants with extremely low birth weight are at risk for intracranial hemorrhage and hypoglycemia resulting from a lack of hepatic glycogen storage. These neonatal problems may have results similar to those of asphyxia. Hyperbilirubinemia may result from increased destruction of red cells (eg, hemolysis due to maternal-child blood group incompatibility) or decreased excretion of bilirubin (eg, due to an immaturity of liver function). The brain damage that may ensue results in manifestations of various degrees, including CP, sensorineural hearing loss, and mental retardation.
Postnatal Causes

Infections

Bacterial and viral infections of the brain during childhood may cause meningitis and encephalitis and result in permanent damage. The number of these complications has decreased because of improved treatment and the availability of immunizations such as that for measles.

Toxic substances

Lead poisoning is still an important cause of mental retardation in the United States. The most frequent source of lead is pica (ie, ingestion of flaking, old, lead-based paint). Other sources of lead are certain fruit-tree sprays, leaded gasoline, some glazed pottery, and fumes from burning automobile batteries. Gastrointestinal symptoms dominate in acute poisoning. Headache may be associated with increased intracranial pressure, which may even lead to coma. Late manifestations include developmental retardation, ataxia, seizures, and personality changes.

Other postnatal causes

Among childhood malignancies, brain tumors are second in frequency after leukemias. Of these, 70-80% are gliomas, symptoms of which depend mostly on location. Some are benign and treatable, but most have deleterious effects, resulting in various neuropsychiatric symptoms depending on their location and extent. In addition, treatment such as surgery and radiation might affect the integrity and function of the brain. Traffic accidents, drowning, and other traumas are the most common causes of death during childhood. Even greater is the number of children who become disabled. Near-drowning is often devastating, but even in these cases, improvement of functional capacity may be achieved by rehabilitation because the developing brain has the ability to recover.

Psychosocial problems

The developmental level of a growing individual depends on the integrity of the CNS and on environmental and psychological factors. The importance of environmental stimulation for child development has been appreciated since research on children in institutions showed that development was severely affected in a depriving environment, even if adequate physical care was provided. Poverty predisposes the child to many developmental risks, such as teenage pregnancies, malnutrition, abuse, poor medical care, and deprivation.

Severe maternal mental illness is another risk factor. Mothers with severe and chronic illness might have difficulty providing adequate care and stimulation. Maternal depression during pregnancy and postpartum has been shown to be associated with developmental delay in children at 18 months of age.[10]

Children of mothers who have schizophrenia are at risk for the development of cognitive deficits, although these may not be secondary to maternal illness but may represent a genetically determined predisposition to schizophrenia. Psychotic illness in a child has been shown to be associated with a decline in cognitive abilities.

Unknown causes

Despite detailed assessment, no cause can be identified in approximately 30% of cases of severe mental retardation and in 50% of cases of mild mental retardation. This, of course, reflects the inadequacy of diagnostic techniques, rather than a lack of causation.
Signs

Children with mental retardation may learn to sit up, to crawl, or to walk later than other children, or they may learn to talk later. Both adults and children with mental retardation may also exhibit the following characteristics:

- Delays in oral language development
- Deficits in memory skills
- Difficulty learning social rules
- Difficulty with problem solving skills
- Delays in the development of adaptive behaviors such as self-help or self-care skills
- Lack of social inhibitors.

The limitations of cognitive functioning will cause a child with mental retardation to learn and develop more slowly than a typical child. Children may take longer to learn language, develop social skills, and take care of their personal needs such as dressing or eating. Learning will take them longer, require more repetition, and skills may need to be adapted to their learning level. Nevertheless, virtually every child is able to learn, develop and become participating members of the community.

In early childhood borderline mental retardation (IQ 71-84) and mild mental retardation (IQ 60–70) may not be obvious, and may not be identified until children begin school. Even when poor academic performance is recognized, it may take expert assessment to distinguish mild mental retardation from learning disability or emotional/behavioral disorders. As individuals with mild mental retardation reach adulthood, many learn to live independently and maintain gainful employment. Moderate mental retardation (IQ 50–60) is nearly always apparent within the first years of life. Children with moderate mental retardation will require considerable supports in school, at home, and in the community in order to participate fully. As adults they may live with their parents, in a supportive group home, or even semi-independently with significant supportive services to help them, for example, manage their finances. A person with severe mental retardation and profound mental retardation below this level will need more intensive support and supervision his or her entire life.

**Intellectual disability / Mental retardation**

Intellectual disability is a condition diagnosed before age 18 that includes below-average intellectual function and a lack of skills necessary for daily living.
In the past, the term mental retardation was used to describe this condition. This term is no longer used.

**Causes**

Intellectual disability affects about 1% to 3% of the population. There are many causes of intellectual disability, but doctors find a specific reason in only 25% of cases.

Risk factors are related to the causes. Causes of intellectual disability can include:

- Infections (present at birth or occurring after birth)
- Chromosomal abnormalities (such as Down syndrome)
- Environmental
- Metabolic (such as hyperbilirubinemia, very high bilirubin levels in babies)
- Nutritional (such as malnutrition)
- Toxic ([intrauterine](#) exposure to [alcohol](#), cocaine, amphetamines, and other drugs)
- Trauma (before and after birth)
- Unexplained (this largest category is for unexplained occurrences of intellectual disability)

**Symptoms**

As a family, you may suspect your child has an intellectual disability when your child has any of the following:

- Lack of or slow development of motor skills, language skills, and self-help skills, especially when compared to peers
- Failure to grow intellectually or continued infant-like behavior
- Lack of curiosity
- Problems keeping up in school
- Failure to adapt (adjust to new situations)
- Difficulty understanding and following social rules

Signs of intellectual disability can range from mild to severe.

**Exams and Tests**

Developmental tests are often used to assess the child:

- Abnormal Denver developmental screening test
- Adaptive Behavior score below average
- Development way below that of peers
- Intelligence quotient (IQ) score below 70 on a standardized IQ test

**Treatment**
Goal of treatment is to develop the person’s potential to the fullest. Special education and training may begin as early as infancy. This includes social skills to help the person function as normally as possible.

It is important for a specialist to evaluate the person for other physical and mental health problems. Persons with intellectual disability are often helped with behavioral counseling.

Discuss your child’s treatment and support options with your health care provider or social worker so that you can help your child reach his or her full potential.

**Outlook (Prognosis)**

Outcome depends on:

- Severity and cause of the intellectual disability
- Other conditions
- Treatment and therapies

Many people lead productive lives and learn to function on their own. Others need a structured environment to be most successful.

**When to Contact a Medical Professional**

Call your health care provider if:

- You have any concerns about your child’s development
- You notice that your child’s motor or language skills are not developing normally
- Your child has other disorders that need treatment

**Prevention**

**Genetic:** Genetic counseling and screening during pregnancy can help parents understand risks and make plans and decisions.

**Social:** Nutrition programs can reduce disability associated with malnutrition. Early intervention in situations involving abuse and poverty will also help.

**Toxic:** Preventing exposure to lead, mercury, and other toxins reduces the risk of disability. Teaching women about the risks of alcohol and drugs during pregnancy can also help reduce risk.

**Infectious diseases:** Certain infections can lead to intellectual disability. Preventing these diseases reduces the risk. For example, rubella syndrome can be prevented through vaccination. Avoiding exposure to cat feces that can cause toxoplasmosis during pregnancy helps reduce disability from this infection.
Alternative Names
Mental retardation

Support Groups
http://www.nichcy.org

http://kidshealth.org/kid/grow/Problems Learning and Functioning
http://www.healthline.com/symptom/mental-retardation

http://www.aaidd.org

The Arc http://www.thearc.org

http://www.nads.org