

# Intellectual Disability in Children — Signs and Treatments

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**Intellectual disability in children refers to a chronic condition of the developmental period involving the limitation in mental functioning and skills such as communication, taking care of oneself and others, etc. Intellectual disability results from any process that either limits the growth of the brain and maturation of cognition or any processes that are toxic to the brain and damage it. These include intrauterine growth restriction, toxin exposure, infections, and placental insufficiencies. Perinatal and postnatal factors such as hypoxia, hyperbilirubinemia, and infections also cause mental retardation.**



## Definition of Intellectual Disability

It is **also known as a cognitive disability or mental retardation** and refers to a chronic condition involving the limitation in mental functioning and skills such as communication, taking care of oneself and others.

This results in social segregation of a child—and thus delayed development of the child's behavior. The onset of sub-average intelligence is usually in the childhood period, therefore, the development of the child is impaired.

## Epidemiology of Intellectual Disability

Intellectual disability among children is one of the most common causes of

developmental disability. **It affects 1-3% of the worldwide population;** all of them require special care but very few receive it. Out of this number, 40–50% have emotional and behavioral problems or are affected by a psychiatric disorder in their lifetime.

**Note:** In the United States of America, 6.5 million children have an intellectual disability, with more than 540000 children requiring special care—such as special education in community-based care centers.

## Etiology and Pathophysiology of Intellectual Disability

Intellectual disability is thought to follow certain triggers that alter the development of the child's brain and cognitive function in uterine life or after birth.

### Genetic mutations/alteration in embryonic development

**Fetal abnormalities such as NTDs are genetically determined,** and the presence of such defects leads to the compromise of brain maturation and cognitive development thus leading to intellectual disability.

**Some genetic conditions** are associated with impaired intellectual ability:

#### Non-inherited conditions

- **Down syndrome:** results from nondisjunction in meiosis II that leads to trisomy 21. It manifests as patients with low intellectual ability and recognizable physical features.
- **Cri-du-chat syndrome:** results from the deletion of one pair of genes on chromosome 5p3.

#### Inherited conditions

**Tuberous sclerosis** is inherited in an autosomal dominant pattern and **results from a mutation of the gene that forms the ectoderm.** Lack of expression of this gene hinders the development of the derivatives of the ectoderm which include nervous tissue, thus leading to intellectual disability. Skin manifestations of angiofibroma and macules are also seen.

**Phenylketonuria** is an autosomal recessive disease characterized by a defective enzyme used in the breakdown of phenylalanine, thus the amino acid accumulates in the body. The chemical is toxic to the brain, and accumulation in developmental stages of life leads to cognitive impairment. **It has associated symptoms of hypertonia, hyperreflexia, musty color of urine and sweat.** Seizures and tremors may also occur in extreme nervous system damage.

**Fragile X syndrome** is an X linked inherited disorder that is characterized by constriction of the X chromosome at Xq27.3 which renders the X chromosome fragile, and it breaks off easily. It **is more common in males and presents with delayed speech development,** restlessness, hyperactivity, inattentiveness, social withdrawal and depression.

## Intrauterine infections

Infections occurring in the uterus can lead to intellectual disability since they interfere with organogenesis causing mental retardation, microcephaly, hearing, and visual impairment. They include the TORCH complex of organisms.

## Exposure to teratogens

**Once the fetus is exposed to teratogens, such as alcohol, the development process is halted** or takes place in a disorganized manner. Microcephaly, growth retardation, CNS dysfunction, and delayed development result.

## Intrauterine growth restriction

It causes microcephaly and altered development of the whole brain, including cognition, thus leads to intellectual impairment. Since it results from nutrient insufficiency, an ischemic injury may also lead to brain damage.

## Perinatal causes

**Perinatal hypoxia occurring during the birth of the baby leads to ischemic injury to the baby's brain** and damage of the brain regions involved in cognitive functions.

Development of perinatal infections such as HSV-2 virus leads to the development of encephalitis which leads to neurological deficits if untreated. This causes profound retardation and microcephaly.

**Hyperbilirubinemia**, which is a common occurrence in the neonatal period, **is toxic to the brain cells and causes damage of the brain, sensorineural hearing loss** and mental retardation. Blood group incompatibility and immature liver function are the most common sources.

## Classification of Intellectual Disability

The diagnostic and statistical manual of mental health classifies intellectual disability into the **following subgroups based on intellectual functioning assessment (IQ score)** in relation to the general population's mean score of 100.

	Intellectual functioning assessment (IQ score)	SD from mean score of the population
<b>Mild Intellectual Disability</b>	50–69	2–3 SDs
<b>Moderate Intellectual Disability</b>	35–49	3–4 SDs
<b>Severe Intellectual Disability</b>	20–34	4–5 SDs
<b>Profound Intellectual Disability</b>	< 20	> 5 SDs

## Clinical features of Intellectual Disability

Clinical features of the disease are seen if the patient is severely incapacitated. In most circumstances, however, **the symptoms manifest once the child begins to attend a school or play with fellow children**. The symptoms that are evident include delay of

motor functions and social development.

## Gross motor

There is delayed achievement of the developmental milestones in comparison to other children. It occurs in association with fine motor delay and infrequently accompanies the cognitive and language delay. Subtle delays in a gross motor acquisition, or clumsiness, may be identified in the developmental assessment.

## Language delay

Affected children usually have a delay in expressive language (speech) and receptive language (understanding).

## Fine motor/adaptive delay

The **children are not able to undertake activities such as self-feeding, toileting and dressing** as seen with the normal population. Prolonged, messy finger feeding and drooling are signs of oral-motor incoordination.

## Cognitive delay

Children with intellectual disability have **difficulties with memory, problem-solving and logical reasoning**. They have difficulty following instructions or directions well displayed in class. Special attention is needed to help them.

## Social delays

Children with MR may display a lack of interest in age-appropriate toys, delays in imaginative play and lack of interest in playing together with other children thus they become social misfits.

## Neurologic and physical abnormalities

Intellectual disability occurs in an array of different conditions and the features commonly associated with it include microcephaly, macrocephaly, history of intrauterine growth retardation, prematurity and congenital anomalies.

## Physical examination findings

### Head circumference

- **Microcephaly**—indicated by lower head circumference for the respective age—correlates highly with cognitive deficits.
- **Macrocephaly** is characterized by a head circumference > 98th percentile of the chart circumference. It may indicate hydrocephalus and is associated with some inborn errors of metabolism.

### Height

These patients feed poorly and **end up having stunted growth with a short stature** that may suggest a genetic disorder, fetal alcohol syndrome or hypothyroidism.

### Neurologic

This examination should include **assessments of head growth, muscle tone, strength and coordination**, deep tendon reflexes, persistent primitive reflexes, ataxia and other abnormal movements such as dystonia.

### **Sensory**

**Vision and hearing should always be tested** particularly among those with severe impairments since these patients have auditory and visual impairments in association with the intellectual disability.

### **Skin**

Cutaneous findings of etiologic interest include hyperpigmented and hypopigmented macules, ash-leaf spots (seen in tuberous sclerosis), fibromas and irregular pigmentation patterns. The skin lesions may indicate the causative pathway or associated malformation.

## **Assess the patient's behavior**

### **Social environmental control**

If the patient has any form of aggressive or self-injurious behavior the treatment of choice is to reinforce the evasion mechanisms.

### **Communication**

It should be determined whether the patient has any problems with attention and communicating with people or following instructions.

### **Modulation of physical discomfort**

Medical conditions must be checked out as they could be the cause of the discomfort.

### **Modulation of emotional discomfort**

These patients are predisposed to the development of certain depressive and bipolar disorders due to the social alienation. They must be identified if present and treated appropriately.

## **Investigations of Intellectual Disability**

Intellectual disability is a **clinical diagnosis based on the physical examination findings** and history taken. The common differential diagnosis and associated conditions are also diagnosed clinically. Therefore, investigations are rarely needed in the diagnosis of intellectual disability. However, some associated congenital anomalies may need a few investigations to confirm their diagnosis.

To ascertain that the patient has an intellectual disability, the following must be done:

### **Assessment of intellectual functioning (IQ score)**

This is performed using standard tests, such as the Binet intelligence testing. The normal population has a mean score of 100. Intellectual disability is defined as a variation of more than 2SDs (30) below the average population score which is a score of < 70.

## Assessment of adaptive behavior and play

Interaction and behavior in comparison with other children of a similar age to demonstrate delay or altered behavior.

## Identification of the physical characteristics

Characteristics that are seen in the diseases associated with intellectual disability such as:

- **Down syndrome:** low set ears, Crimean crease, congenital heart lesions;
- **Phenylketonuria:** hypertonia, hyperreflexia, seizures, musty color of urine and sweat;
- **Fragile X syndrome:** restless hyperactive males.

Some investigations to consider include:

<b>Genetic Testing</b>	<ul style="list-style-type: none"><li>• Helps to establish chromosomal abnormalities in the inherited causes of intellectual disability.</li></ul>
<b>Metabolic Testing</b>	<ul style="list-style-type: none"><li>• Tests such as thyroid function tests and phenylalanine levels are used to determine the cause of brain damage and mental retardation.</li></ul>
<b>EEG</b>	<ul style="list-style-type: none"><li>• Used in the assessment of seizure episodes that may be seen in phenylalanine- and bilirubin-induced damage of the brain.</li></ul>
<b>Blood workups (complete blood count, renal and liver function tests)</b>	<ul style="list-style-type: none"><li>• May be needed in ruling out systemic causes of mental retardation such as hyperbilirubinemia.</li><li>• They are needed for preparation of the patient for other tests and procedures such as contrast enhanced imaging.</li></ul>
<b>Brain Imaging</b>	<ul style="list-style-type: none"><li>• MRI and CT scans of the brain are the gold standard investigations for demonstration of cerebral dysgenesis and calcifications within the brain.</li></ul>

## Differential Diagnosis of Intellectual Disability

<b>Microcephaly</b>	<ul style="list-style-type: none"><li>• A small for respective age head circumference that indicates severe underlying cognitive deficits.</li></ul>
<b>Major depressive disorder</b>	<ul style="list-style-type: none"><li>• Associated with low mood and may be evident in patients with mental retardation.</li></ul>
<b>Cretinism</b>	<ul style="list-style-type: none"><li>• Have associated general retardation of growth in addition to mental retardation.</li></ul>
<b>Substance abuse disorder</b>	<ul style="list-style-type: none"><li>• Accompanied by history of the substance use and other symptoms, such as psychosis, that are related to substance use in addition to the brain damage that may occur in early childhood use.</li></ul>

## Management of Intellectual Disability

Mental retardation is **treated mainly by psychosocial counseling and environmental modulation** to avoid the occurrence of depression and other mental illnesses. Once they occur psycho-pharmacology should be employed.

### Psychosocial therapy

This is applied to **address behavioral problems:**

Behavioral interventions such as behavioral analysis and reward of specific behavior at their occurrence lead to an acceleration of the rewarded behavior. Behavior decelerating tactics involve rewarding in the period bad behavior did not occur.

Environmental **management and elimination of any stresses or triggers of bad behavior** such as noise, increased temperature, the transition from school or residence, personal loss or rejection by friends, hostility from friends, physical abuse and

confrontation of large crowds.

Client and family education is to allow the patient to get maximum attention and support in his/her activities. This reduces the social alienation associated with mental retardation and the accompanying occurrence of depressive episodes.

## Psycho-pharmacological therapy

The use of pharmacological methods **is restricted only to the diagnosis that is made as per the DSM V criteria**. Clozapine and risperidone are common medications for depression and behavior modulation.

## Other forms of therapy

The doctor-patient relationship is important in treating and preventing the occurrence of depressive episodes in both the affected child and his/her parents. **Genetic counseling**, especially when a prenatal diagnosis is made, is necessary. This treatment can be instituted together with other therapies.

**Nutritional therapy** such as intake of a low phenylalanine diet soon after birth is useful to avoid brain damage upon identification of PKU. The dietary restriction is continued for life, although the most important period is the developmental period.

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