

# INTELLECTUAL DISABILITY

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Suggested citation: Ke X, Liu J. Intellectual disability. In Rey JM (ed), *IACAPAP e-Textbook of Child and Adolescent Mental Health*. Geneva: International Association for Child and Adolescent Psychiatry and Allied Professions 2012.

The term *intellectual disability* (ID) is increasingly being used instead of *mental retardation*. ID or mental retardation is defined as *a condition of arrested or incomplete development of the mind, which is especially characterized by impairment of skills manifested during the developmental period, which contribute to the overall level of intelligence, i.e., cognitive, language, motor, and social abilities* (World Health Organization, WHO, 1992). The American Association on Intellectual and Developmental Disabilities (AAIDD) describes ID as *characterized by significant limitations both in intellectual functioning and in adaptive behavior as expressed in conceptual, social, and practical adaptive skills*. This disability originates before age 18. In general, ID applies to the same individuals who were previously diagnosed with mental retardation in kind, level, type, duration and the need for services and supports. Every individual who is or was eligible for a diagnosis of mental retardation is eligible for a diagnosis of ID (Schalock et al, 2007).

## EPIDEMIOLOGY

There have been many surveys to ascertain the prevalence of ID across the world with estimates ranging from 1% to 3% (Harris, 2006). A recent meta-analysis concluded that the average prevalence of intellectual disability across all the studies is 1%. Prevalence is higher in males in both adult and child and adolescent populations. Among adults, the female-to-male ratio varies between 0.7:1 and 0.9:1, while in children and adolescents it ranges between 0.4:1 and 1:1. Rates vary according to income; the highest prevalence occurs in low and middle income countries where rates are almost twice those in high income countries (Maulik et al, 2011). Another meta-analysis, which considered studies published between 1980 and 2009 in European countries, found overall estimates ranging from 0.4% and 1.4% (Wittchen et al, 2011). The prevalence of ID across Asia is broadly consistent with estimates in western countries: 0.06%-1.3% (Jeevanandam, 2009). The most recent Chinese national survey on disability, conducted in 2006, estimated a prevalence of ID of 0.75%. Prevalence in urban areas was lower (0.4%) than in rural areas (1.02%) (Kwok et al, 2011)

## ETIOLOGY AND RISK FACTORS

Etiology of ID is heterogeneous. Injury, infections and toxins have become less prevalent causes because of improved antenatal care, while genetic factors have become more prominent. No specific etiology can be found in up to 40% of cases, particularly in mild ID. Environmental influences (e.g., malnutrition, emotional and social deprivation experienced, for example, in poorly run orphanages) can also cause or aggravate ID. Understanding the etiology of ID raises the possibility of treatment or prevention in some cases, while it may allow predicting specific difficulties in others.

Many factors have been confirmed to cause or be associated with ID. These factors, which influence the development and function of the child's brain prenatally, perinatally or postnatally, can be divided into three groups: organic, genetic and socio-cultural. Trisomy 21 and fragile X are the commonest diagnosable genetic causes of intellectual disability. It is unlikely that all intellectual disability will fit neatly into these three groups – overlapping genetic, environmental and socio-cultural factors are likely to be relevant in many cases. Conversely, in up to two-thirds of mild cases and one-third of severe cases, no causes are found,



### What is a normal development?

There is a wide range of what can be considered "normal" but growth follows a certain sequence. Particular skills are expected to emerge at more or less a specific age. For a detailed description of normal developmental milestones go to Chapter A.2 or click on the picture to go to the National Dissemination Center for Children with Disabilities .

**Table C.1.1 Common causes of intellectual disability**

Category	Type	Examples
Prenatal (before birth)	Chromosomal disorders	<ul style="list-style-type: none"> <li>Down's syndrome*</li> <li>Fragile X syndrome</li> <li>Prader Willi syndrome</li> <li>Klinefelter's syndrome</li> </ul>
	Single gene disorders	<ul style="list-style-type: none"> <li>Inborn errors of metabolism, such as galactosemia*</li> <li>Phenylketonuria*</li> <li>Mucopolysaccharidoses</li> <li>Hypothyroidism*</li> <li>Tay-Sachs disease</li> <li>Neuro-cutaneous syndromes such as tuberous sclerosis and neurofibromatosis</li> <li>Brain malformations such as genetic microcephaly, hydrocephalus and myelo-meningocele*</li> <li>Other dysmorphic syndromes, such as Laurence-Moon-Biedl syndrome</li> </ul>
	Other conditions of genetic origin	<ul style="list-style-type: none"> <li>Rubinstein-Taybi syndrome</li> <li>Cornelia de Lange syndrome</li> </ul>
	Adverse environmental influences	<ul style="list-style-type: none"> <li>Deficiencies* such as iodine deficiency and folic acid deficiency</li> <li>Severe malnutrition in pregnancy*</li> <li>Substances use* such as alcohol (fetal alcohol syndrome), nicotine and cocaine during early pregnancy</li> <li>Exposure* to other harmful chemicals such as pollutants, heavy metals, abortifacients, and harmful medications such as thalidomide, phenytoin and warfarin in early pregnancy</li> <li>Maternal infections such as rubella*, syphilis*, toxoplasmosis, cytomegalovirus and HIV</li> <li>Others, such as excessive exposure to radiation* and Rh incompatibility*</li> </ul>
Perinatal (around the time of birth)	Third trimester (late pregnancy)	<ul style="list-style-type: none"> <li>Complications of pregnancy*</li> <li>Diseases* in mother, such as heart and kidney disease, diabetes</li> <li>Placental dysfunction</li> </ul>
	Labour (during delivery)	<ul style="list-style-type: none"> <li>Severe prematurity, very low birth weight, birth asphyxia</li> <li>Difficult or complicated delivery*</li> <li>Birth trauma*</li> </ul>
	Neonatal (first four weeks of life)	<ul style="list-style-type: none"> <li>Septicemia, severe jaundice*, hypoglycemia</li> </ul>
Postnatal (in infancy and childhood)		<ul style="list-style-type: none"> <li>Brain infections such as tuberculosis, Japanese encephalitis, and bacterial meningitis</li> <li>Head injury*</li> <li>Chronic lead exposure*</li> <li>Severe and prolonged malnutrition*</li> <li>Gross under stimulation*</li> </ul>

\*Definitely or potentially preventable.

highlighting the need for further research. A more detailed list of causes is available at the WHO website (see Table C.1.1). A few are described below in more detail.

### Intelligence quotient (IQ)

IQ is a score derived from one of several tests. There are many types of IQ tests that seek to measure general or specific abilities: reading, arithmetic, vocabulary, memory, general knowledge, visual, verbal, abstract-reasoning etc. Well-known IQ tests include the Wechsler Intelligence Scale for Children, Stanford-Binet, Kaufman Assessment Battery for Children, and Raven's Progressive Matrices. Traditionally, an IQ score was obtained by dividing the mental age of the person taking the test (the age group which on average scored such a result in a random sample of the population) by the chronological age multiplied by 100. However, this method has shortcomings (e.g., it cannot be used in adults). Currently the test results are standardised against a representative sample of the population; IQ scores for children are relative to children of the same age. The median result is defined to be 100 and one standard deviation is 15 points, therefore, 95% of the population have scores within two standard deviations of the mean (i.e., within an IQ range of 70 to 130). For IQ to be accurate needs to be standardised against a population culturally similar to that of the person being tested. For example, using norms obtained in a Brazilian population would produce biased results if the person taking the test is Burmese.

Although IQ can change to some extent with increasing age, it is a surprisingly robust construct that is strongly predictive of achievement. IQ has a large inherited component but environmental factors have a strong effect as well. Heritability increases with increasing age: it can be as low as 0.2 in infancy, 0.4 in middle childhood, and up to 0.8 in adulthood. What appears to be a straightforward concept has been marred by controversy over the years. For example, some scholars believe that intelligence is a learned combination of many different skills and abilities while others assume that intelligence is a single trait that is heavily determined by genetics, even others believe that there are large ethnic or racial differences.

IQ tests are different from achievement tests, the latter seek to measure the skills and knowledge learned (e.g., language, arithmetic), usually through schooling; IQ tests measure aptitude rather than actual achievement (see Chapter C.3). While in the past there was an emphasis on the so-called "general intelligence" current theories view intelligence as a more complex ensemble of aptitudes in a variety of areas (musical, mechanical, physical, social) which can differ substantially in the same individual.

Table C.1.2 illustrates the attainment in adulthood of people with different degrees of ID (WHO). It is clear that even those with severe ID can become at least partly independent in looking after themselves through proper supervision, care and training.

## MANIFESTATIONS AND SUBTYPES

The manifestations of ID are mainly developmental delay in intellectual functioning and deficits in social adaptive functioning. According to the severity of the delay in intellectual functioning, deficits in social adaptive function and IQ, the psychiatric classifications describe four levels of severity:



Hashan is a four year old boy. He still can't walk independently but can take a few steps with support. He recognizes family members but cannot show where his ear and nose are. He can babble (say ba-ba-ba) but has not learnt to say any meaningful word. He can't indicate toileting needs. His parents say that he is like a one-year-old child.

Does Hashan suffer from ID? If so, how severe?

**Table C.1.2 Adult attainment according to the degree of intellectual disability**

Degree	IQ range	Adult attainment
Mild	50-70	<ul style="list-style-type: none"> <li>• Literacy +</li> <li>• Self-help skills ++</li> <li>• Good speech ++</li> <li>• Semi-skilled work +</li> </ul>
Moderate	35-50	<ul style="list-style-type: none"> <li>• Literacy +/-</li> <li>• Self-help skills +</li> <li>• Domestic speech +</li> <li>• Unskilled work with or without supervision +</li> </ul>
Severe	20-35	<ul style="list-style-type: none"> <li>• Assisted self-help skills +</li> <li>• Minimum speech +</li> <li>• Assisted household chores +</li> </ul>
Profound	Less than 20	<ul style="list-style-type: none"> <li>• Speech +/-</li> <li>• Self-help skills +/-</li> </ul>

Note: +/- sometimes attainable; + attainable; ++ definitely attainable

- *Profound*

IQ is usually below 20; profound intellectual disability accounts for 1% to 2% of all cases. These individuals cannot take care of themselves and have no language. Their capacity to express emotions is limited and poorly understood (Adams & Oliver, 2011). Seizures, physical disabilities, and reduced life expectancy are common.

- *Severe*

IQ is usually between 20 and 34; severe intellectual disability accounts for 3% to 4% of all cases. Every aspect of their development in the early years is distinctively delayed; they have difficulty pronouncing words and have a very limited vocabulary. Through considerable practice and time, they may gain basic self-help skills but still need support at school, home and in the community.

- *Moderate*

IQ is usually between 35 and 49, accounting for about 12% of all cases. They are slow in meeting intellectual developmental milestones; their ability to learn and think logically is impaired but are able to communicate and look after themselves with some support. With supervision, they can perform unskilled or semiskilled work.

- *Mild*

IQ is usually between 50 and 69 and account for about 80% of all cases. Development during their early life is slower than in normal children and developmental milestones are delayed. However, they are able to communicate and learn basic skills. Their ability to use abstract concepts, analyze and synthesize are impaired but can achieve reading and computing skills to grade three to six level. They can

perform house-work, look after themselves and do unskilled or semi-skilled work. They usually require some support.

## CLINICAL SYMPTOMS

### Speech

Children with ID usually have delayed language development and difficulties speaking and expressing themselves. The degree of severity varies with the level of impairment of intellectual ability. Mild cases can achieve language skills that are only a little poorer than children in the normal range of development. Severe or profound cases can't communicate at all or speak only a few words.

### Perception

Children with ID are slow in reacting and perceiving environmental stimuli. They have difficulties distinguishing small differences in the shape, size and color.

### Cognition

Capacity to analyze, reason, comprehend and calculate, and for abstract thinking is often impaired to a greater or lesser extent according to severity. Children with mild ID are capable of achieving reading and mathematics skills to approximately the level of a typical child aged 9 to 12 (Daily et al, 2000). Individuals with severe or profound ID lack the capacity to read, calculate or even understand what others say.

### Concentration and memory

Ability to concentrate is low and narrow. By and large, memory is poor and they are slow at remembering although there are exceptions (e.g., savants). They have difficulties recalling and their memories are often inaccurate.

### Emotion

Emotions are often naive and immature but may improve with age. Capacity for self-control is poor and impulsive and aggressive behavior is not uncommon. Some are timid, withdrawn and shy.

### Movement and behavior

Children with ID often lack coordination, may be clumsy or show excessive movement. Meaningless or stereotyped movements (e.g., rocking, head-banging, teeth-biting, shouting, tearing clothes, pulling hair, playing with the genitals) are frequent in severe ID. Destructive, aggressive or violent behavior can also be observed. Self-injurious behavior (e.g. self-slapping or biting) may occur in moderate and severe ID.

### Health problems associated with intellectual disability

Compared with normal children, children with ID are at a higher risk of having other health problems. The most prevalent health conditions are: epilepsy (22%), cerebral palsy (20%), anxiety disorders (17%), oppositional defiant disorder (12%), and autistic disorder (10%) (Oeseburg et al, 2011).

### *Epilepsy*

- Between 1% and 13% of children with Down syndrome have epilepsy (Arya et al, 2011)
- Epilepsy, often severe and hard to control, is present in 85% of

Angelman syndrome patients within the first three years of life (Fiumara et al, 2010)

- Patients with fragile X syndrome are highly prone to develop epilepsy (Qiu et al, 2008)

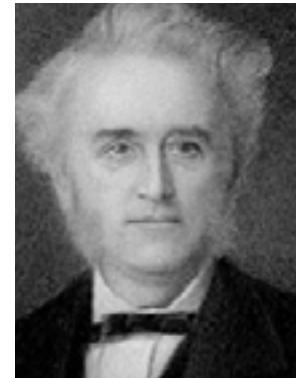
**Behavior problems**

Symptoms like restlessness (continuously moving around, unable to sit in one place), poor concentration, impulsiveness, temper tantrums, irritability and crying are common. Other disturbing behavior, like aggression, self-injurious behavior (such as head banging) and repetitive rocking may also be seen (see section on challenging behaviors below). When such behavior is severe and persistent, it can become a major source of stress for families. Therefore, attention should be paid to reduce such behavior while providing treatment and care.

**Sensory impairment**

Visual and hearing problems are present in about 5%-10% of persons with ID. Sometimes these problems can be resolved by using hearing aids or glasses, or undergoing surgery for cataracts

As noted earlier, other developmental disabilities, such as cerebral palsy, speech problems and autism can occur along with ID. Persons with multiple disabilities pose a big challenge in terms of providing care.



John Langdon Haydon Down (1828 -1896), a British physician, was the first to describe a relatively common genetic disorder that is now called Down syndrome.

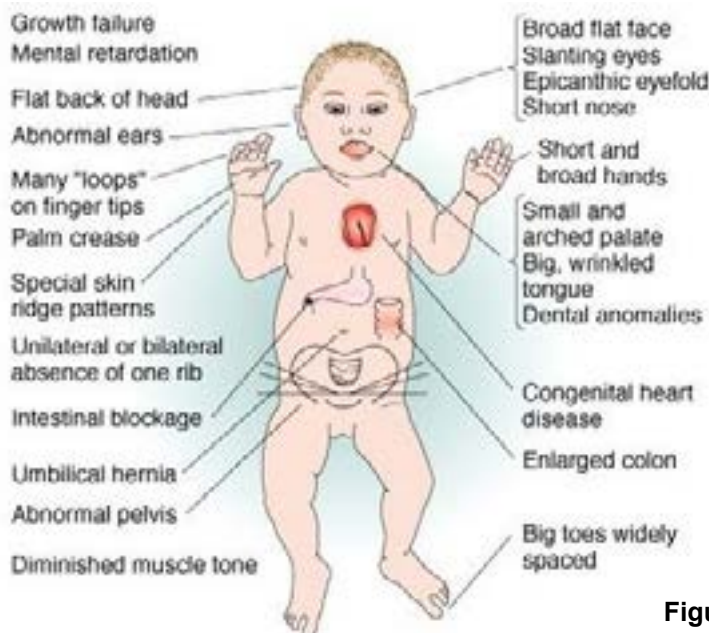
**COMMON CONDITIONS ASSOCIATED WITH INTELLECTUAL DISABILITY**

**Down syndrome**

Down syndrome, also known as trisomy 21, is a chromosomal disorder caused by an additional copy of genetic material on chromosome 21, which affects the development of the body and brain. This syndrome was first described by the British physician John Langdon Down and identified in 1959 as caused by a 21 trisomy by Jérôme Lejeune.

**Maternal age and Down syndrome**  
 A woman's risk of having a baby with Down syndrome is:

- At age 25, 1 in 1,250
- At 30, 1 in 1,000
- At 35, 1 in 400
- At 40, 1 in 100
- At 45, 1 in 30
- At 49, 1 in 10



**Figure C.1.1 Symptoms of Down syndrome**

The incidence of Down syndrome is approximately one per 1000 newborns (Roizen & Patterson, 2003) and is influenced by maternal age. Women aged 35 and older have significantly higher risk.

Down syndrome can be diagnosed by chromosome analysis either prenatally or postnatally, according to which it can be grouped into four types: trisomy 21, mosaicism, translocation and duplication of a portion of chromosome 21.

Clinical features of Down syndrome include (Figure C.1.1):

- Intellectual disability, usually mild; they possess good social skills
- A characteristic appearance including brachycephaly, epicanthal folds, upslanting palpebral fissures, strabismus, Brushfield spots on iris, flattened nose, low-set and rounded ear, macroglossia, open mouth, short neck, brachydactyly, fifth finger clinodactyly, atypical fingerprints, wide 1-2 toe gap known as sandal foot
- Impaired physical growth such as short stature, short limbs and lax ligaments
- Often accompanied by different medical problems including congenital heart disease, duodenal atresia, hearing loss, ophthalmological problems, hypothyroidism, early-onset dementia, and leukemia.

Down syndrome can be detected through prenatal screening. Common screening procedures include: (a) measurement of maternal serum alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG), unconjugated oestriol, and inhibin-Alpha (INHA) at 15-20 gestational weeks; (b) fetal ultrasound testing for a thickened nuchal fold with measurement of maternal serum free Beta hCG and pregnancy-associated plasma protein A (PAPPA) at 10-13.5 gestational weeks; both (a) and (b). For families with a high risk of having a child with Down syndrome, an invasive diagnostic test, such as amniocentesis, chorionic-villus sampling, or percutaneous umbilical cord blood sampling, performed in the late first trimester or early second trimester, is most accurate.

### Fragile X syndrome

Fragile X syndrome (also known as Martin-Bell syndrome and Escalante's syndrome) is an X-linked disease that is one of the most common inherited forms of ID. It is also associated with autism. Martin and Bell first described this disorder in 1943, and Herbert Lubs identified an associated fragile site on the X chromosome in 1969.

The fragile X syndrome is characterized by an expansion of a single trinucleotide gene sequence to over 200 copies of a CGG repeat in the 5'-untranslated region of the fragile X ID 1 (FMR1) gene located at band q27.3 on the long arm of the X chromosome (Xq27.3), which silences the transcription of the gene.

Incidence is about 1 per 2000-5000 persons and is 30% more frequent in males than in females (Hessl et al, 2002; Ridaura-Ruiz et al, 2009). Fragile X syndrome is an X-linked dominant condition with variable expressivity and possibly reduced penetrance that is largely transmitted by females but affecting males more often because males normally have only one copy of the X-chromosome.

### Premutation

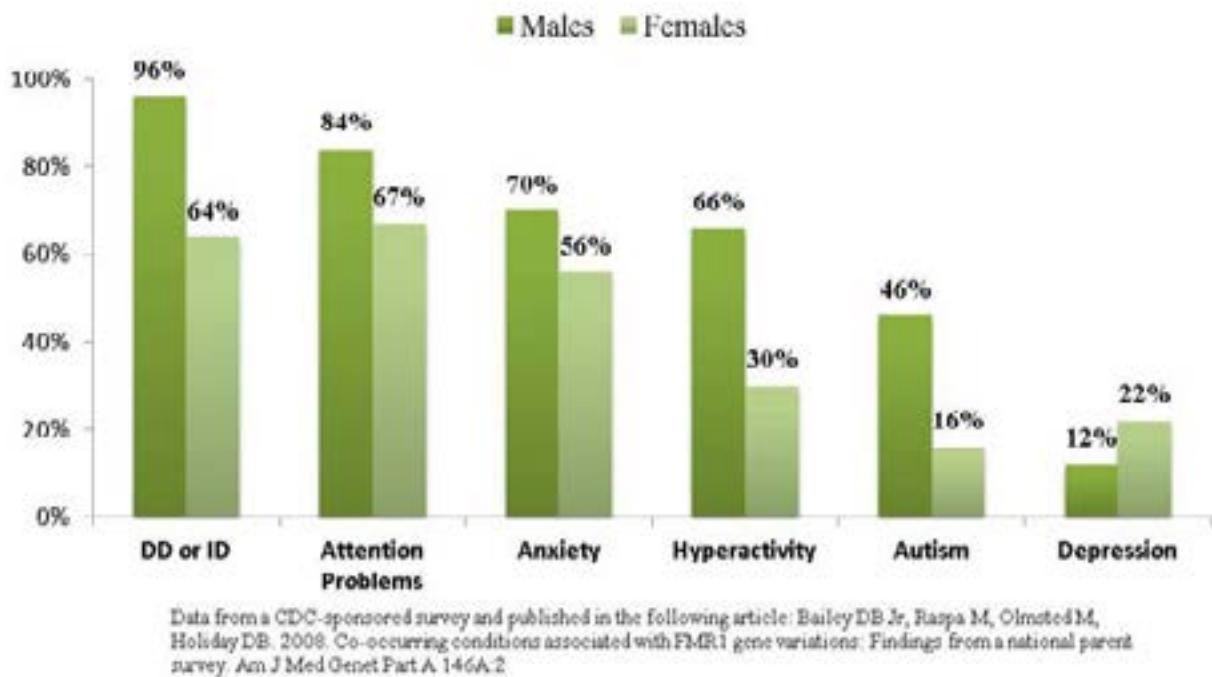
Premutation is a change in a gene that precedes a mutation without altering the function of the gene. In disorders caused by trinucleotide repeat expansions, such as fragile X, premutation is an abnormally large allele that is not associated with clinical symptoms but that can expand into a full mutation when transmitted to the offspring. Full mutations cause clinical symptoms of the disorder.

### Prevalence of fragile X

Approximately:

- 1 in 3600 to 4000 males are born with the full mutation for fragile X and will have fragile X syndrome
- 1 in 4000 to 6000 females are born with the full mutation for fragile X but only 50% of them will show some features of fragile X syndrome
- 1 in 800 men are carriers of the fragile X premutation
- 1 in 260 women are carriers of the fragile X premutation.



**Figure C.1.2 Percentage of children with fragile X syndrome diagnosed or treated for other conditions**

Source: CDC

Clinical manifestations of the fragile X syndrome vary from mild to severe in physical, cognitive, emotional and behavioral features. Generally, females have a less severe form of the disease than males. Physical phenotype includes a long, narrow face with a prominent forehead and protuberant ears, joint hypermobility associated with connective tissue dysplasia, double-jointed thumbs, flat feet, and macro-orchidism after puberty in males. Individuals with fragile X syndrome usually have cognitive deficits – IQ ranging from normal or borderline to severely low – such as problems with working memory, executive function, and mathematic and visuospatial abilities. Language delay is also evident in early childhood. Emotional and behavioral disturbances are common, including anxiety and mood disorders, features of attention deficit hyperactivity disorder, obsessive compulsive-like symptoms (e.g., repetitive actions or phrases), aggressive and self-injurious behavior, and a difficult temperament. Fragile X syndrome is a common cause of autism. Neurological problems such as seizures may also be found. In addition, persons carrying a premutation (CGG repeat number ranges from 55 to 200) are believed to have a clinical disorder characterized by mild learning disability, emotional problems, premature ovarian failure, and a neurodegenerative disorder called fragile X-associated tremor/ataxia syndrome in older people.

Genetic tests for CGG repeat expansions in FMR1 gene using PCR and southern blot analysis is diagnostic for fragile X syndrome and should be provided to all persons with developmental delay, borderline intellectual abilities, ID and autism. Sequencing of FMR1 gene should be considered as well to exclude deletions of this gene if normal CGG-repeat length is found (Garber et al, 2008).



Prominent characteristics of Fragile X syndrome include an elongated face, large or protruding ears, and low muscle tone.

Genetic counseling is recommended for the whole family if a positive fragile X syndrome premutation or full mutation is detected, and cascade testing should be planned for family members. Although genetic counseling cannot prevent fragile X syndrome, it is still important to give at-risk families accurate reproductive counseling and allow for appropriate intervention beginning in infancy.

### Phenylketonuria

Phenylketonuria (PKU) is an autosomal recessive single gene disorder discovered by the Norwegian physician Ivar Asbjørn Følling in 1934. It is caused by mutations of the phenylalanine hydroxylase (PAH) gene or genes coding for enzymes involved in the cofactor tetrahydrobiopterin (BH4) biosynthesis or recycling, which result in dysfunction of phenylalanine metabolism leading to excessive phenylalanine and related substances in the blood, the brain and the urine. Increased phenylalanine concentrations in the brain are toxic and cause disruption of neuropsychological function. The prevalence of phenylketonuria varies widely around the world due to ethnic and social reasons (e.g., frequency of consanguinity). In Europe the prevalence is about one case per 10000 live births, but it is one in 4000 in Turkey, one in 25000 to one in 50000 in Latin America and one in 100000 live births in some regions of China (Blau et al, 2010).

PKU is diagnosed if blood amino acid analysis reveal a raised concentration of phenylalanine (>120  $\mu\text{mol/L}$ ). To clarify whether the patient with hyperphenylalaninaemia is deficient in BH4 synthesis or regeneration, measurement of urinary pterins or red blood cell dihydropteridine reductase through a filter paper-dried blood specimen, or BH4-load test should be done (Blau et al, 2005). Urinary pterins patterns can differentiate several types of PKU:

- *Classical PKU with PAH deficiency*: total pterins are high but the ratio between neopterin and biopterin is normal
- *GTP cyclohydrolase (GTP-CH) I deficiency*: total biopterins are very low or not detectable
- *6-pyruvoyl-tetrahydropterin synthase (6-PTS) deficiency*: neopterin is increased but biopterin is decreased
- *Pterin-4 $\alpha$ -carbinolamine dehydratase deficiency*: neopterin is high while biopterin is low or borderline, and primapterin is high
- *Dihydropteridine reductase (DHPR) deficiency*: neopterin is normal and biopterin is raised.



Click on the picture to view a short description of PKU (6:02)

### Inherited metabolic disorders

Chromosomes in human cells contain genes which control growth and maturation. Some of these are responsible for chemical (metabolic) reactions, which are essential for brain growth. If such a gene is abnormal, it can lead to a derangement of metabolic reactions and cause problems such as mental retardation. Phenylketonuria is one such condition. Babies with phenylketonuria, in addition to mental retardation, have light-colored hair and skin, a small head, and are prone to convulsions.

Blood is taken from a two-week old infant to test for PKU  
Photo: Nando Peretti Foundation

Babies with PKU seem normal at birth but progressively show developmental disabilities frequently accompanied by lighter skin, hair, eyes, eczematous rash, “mousy” odor, motor deficits, seizures, behavior problems and autism. Early screening, diagnosis and prompt intervention can prevent individuals with PKU from further damage to the brain. Newborn blood screening test for PKU is usually performed three to seven days after birth and a repeat test at approximately two weeks of age to verify the initial test, followed by further diagnostic tests if positive.

Intervention should be started as soon as PKU is confirmed. Restriction of dietary phenylalanine is still the most important and effective management: phenylalanine-free formula with low protein; avoiding foods rich in protein (such as meat, fish, milk, eggs, standard bread, most cheeses, nuts and seeds) and containing aspartame (flour, soya). For patients with the BH<sub>4</sub>-responsive type, identified by BH<sub>4</sub>-load test, preparations containing BH<sub>4</sub> may be prescribed. Other approaches, such as large neutral amino acids treatment, use of phenylalanine ammonia lyase and gene therapy are under investigation (Blau et al, 2010). For best outcomes, serum phenylalanine should be monitored regularly throughout life in case changes are needed to maintain target therapeutic blood levels.

### **Congenital hypothyroidism**

Congenital hypothyroidism is an endocrine disease caused by thyroid hormone deficiency after birth. It can be classified into permanent and transient. Permanent congenital hypothyroidism is usually associated with thyroid dysgenesis, dysfunction of thyroid hormone biosynthesis or metabolism, or deficiency of thyroid stimulating hormone (TSH), while transient congenital hypothyroidism is due to iodine deficiency, maternal intake of antithyroid drugs, or transplacental maternal thyrotropin receptor blocking antibodies (TRB-Ab). Congenital hypothyroidism occurs in approximately one per 2000 to 4000 live births (Rastog et al, 2010).

Congenital hypothyroidism is diagnosed by measuring serum TSH and either free T<sub>4</sub> or total T<sub>4</sub> combined with T<sub>3</sub> resin uptake. According to age-normative reference ranges, primary congenital hypothyroidism is confirmed with increased TSH and decreased or normal free T<sub>4</sub> or total T<sub>4</sub>, while secondary (central) congenital hypothyroidism is likely if T<sub>4</sub> is low but TSH is not elevated. There are other diagnostic tests to further determine the underlying etiology including measurement of urinary iodine, radionuclide uptake and scan, thyroid ultrasonography, serum thyroglobulin measurement, antithyroid antibody determinations, evaluation for other pituitary hormone deficiencies, brain MRI, and genetic testing.

Clinical features of congenital hypothyroidism in infants include: persistent jaundice, poor feeding, quiet, excessive sleeping, constipation, low body temperature, abnormal cry, umbilical hernia, bradycardia, hypotonia with delayed reflexes. Some may have a palpable goiter. The typical appearance includes a wide posterior fontanel, puffy face, flattened nose, eyes exhibiting pseudohypertelorism, and open mouth with macroglossia. If untreated, congenital hypothyroidism can result in growth failure, permanent mental impairment and cardiac problems.

Newborn thyroid screening tests and early management are very important to prevent the development of ID in congenital hypothyroidism. Blood sampled from a heel-prick between two and five days of age is screened by special filter

Iodine deficiency (which causes hypothyroidism) is the single greatest preventable cause of mental retardation and an important public health problem. This condition can be prevented with iodine supplements in the diet such as by using iodized salt.

paper cards to detect TSH levels. A second test is performed, especially for preterm and acutely ill term infants with “delayed TSH rise”, between two and six weeks of age. If initial TSH  $>30\text{mU/L}$  serum or  $>15\text{mU/L}$  whole blood, confirmatory serum thyroid testing should be performed (Rastog & LaFranchi, 2010).

Once congenital hypothyroidism is diagnosed, oral thyroxine treatment should be initiated immediately with close follow up, particularly in the first two to three years of life, crucial for positive neurologic outcome. High dose levothyroxine is recommended to normalize serum  $T_4$  and TSH as rapidly as possible, monitoring closely in order to adjust levothyroxine dose promptly if results are abnormal. For infants with central congenital hypothyroidism, however, low dose levothyroxine should be started and increased slowly, with addition of physiological doses of cortical hormone meanwhile to prevent sudden hypocorticism from happening. Treatment of transient congenital hypothyroidism takes several years, whereas it is lifelong in permanent congenital hypothyroidism. Genetic counseling and antenatal diagnosis should be considered for families at risk of having a baby with congenital hypothyroidism.

### Prader-Willi syndrome

Prader-Willi syndrome is an uncommon genetic disorder of chromosome 15q11-13. It was first described by Andrea Prader and Heinrich Willi in 1956. Symptoms include weak muscle tone, feeding difficulties, short stature, incomplete sexual development, cognitive disabilities, and a chronic feeling of hunger that can lead to excessive eating and obesity. Incidence is approximately one in 25,000 to one in 1,000 newborns (Killeen, 2004). Individuals with Prader-Willi syndrome are at risk of learning and attention difficulties. Research suggests that most (50%-



The children depicted here have a genetically confirmed diagnosis of Angelman syndrome. Happy expression and an unstable gait accompanied by uplifted arms are commonly observed. At times, the facial appearance can suggest the diagnosis, but usually facial features are not distinctive. Source: [Gene Reviews](#).

65%) fall within the mild/borderline/low average intelligence range (Curfs & Fryns, 1992; Cassidy, 1997).

Traditionally, Prader-Willi syndrome was diagnosed by clinical characteristics but it can now be diagnosed by genetic testing. Prader-Willi syndrome has no cure. Early diagnosis allows for early intervention. Children should receive treatment to improve muscle tone. Speech and occupational therapy are also indicated. School aged children will benefit from a highly structured learning environment as well as special education. Daily recombinant growth hormone injections are helpful (Carrel et al, 2002).

### **Angelman Syndrome**

Angelman syndrome is a complex genetic disorder characterized by intellectual and developmental delay, severe speech impairment, seizures, ataxia, hand-flapping, and a happy, excitable demeanor with frequent smiling and laughter. It was first described by Harry Angelman in 1965. Prevalence is approximately one in 10,000 to one in 20,000 live births (Petersen et al, 1995; Steffenburg et al, 1996).

Angelman syndrome is caused by the loss of the normal maternal contribution to a region of chromosome 15, most commonly by deletion of a segment of that chromosome. Diagnosis relies on a combination of clinical features, molecular genetic testing or cytogenetic analysis. Consensus diagnostic criteria for Angelman syndrome are available (Williams, 2006). Analysis of parent-specific DNA methylation imprints in the 15q11.2-q13 chromosome region detects approximately 78% of individuals with Angelman syndrome; fewer than 1% have a cytogenetically visible chromosome rearrangement. UBE3A sequence analysis detects mutations in an additional 11%. Accordingly, molecular genetic testing identifies alterations in approximately 90% of individuals with Angelman syndrome (Dagli & Williams, 2011). Currently, Angelman syndrome has no cure; treatment is symptomatic (e.g., epilepsy can be controlled with anticonvulsant medication).

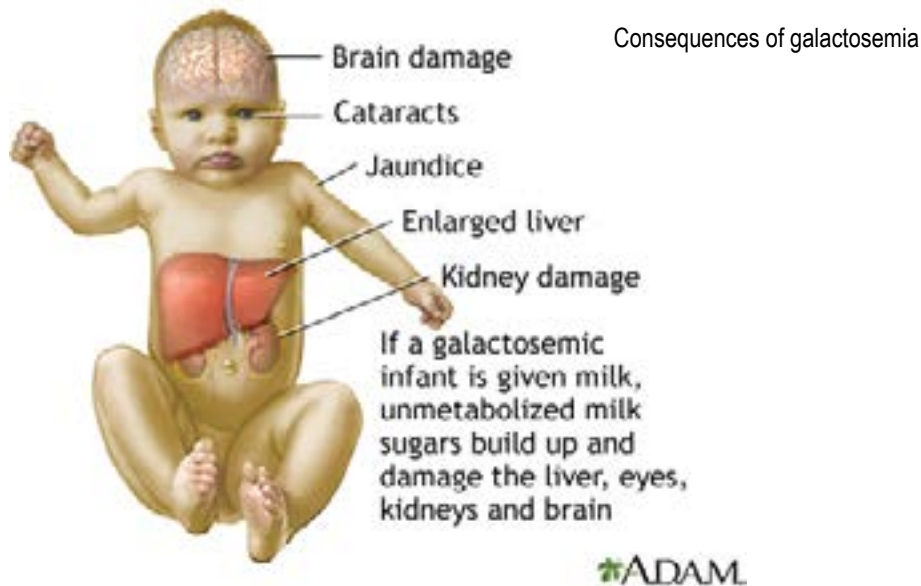
### **Galactosemia**

Galactosemia is an autosomal recessive single gene disorder associated with a dysfunction of the enzymes that convert galactose into glucose, leading to the accumulation of toxic amounts of galactose in the blood and body tissues, resulting in ID and multiple organ damage. It was first reported by Goppert in 1917, and identified as a defect of galactose metabolism by Herman Kalckar in 1956. Its prevalence is about one per 60,000 live births.

- According to the enzymes affected, galactosemia can be classified into three types: *Type I, classic galactosemia*, is due to galactose-1-phosphate uridylyltransferase (GALT) deficiency
- *Type II, galactokinase (GLK) deficiency*
- *Type III, UDP-galactose epimerase (GALE) deficiency*.

Diagnosis of galactosemia is established by a test using blood or urine to detect activity of the three enzymes mentioned above and to quantify galactose levels. In addition, molecular genetic testing is now available (Elsas, 2010).

Infants with galactosemia present nonspecific symptoms including vomiting, diarrhea, poor feeding, prolonged jaundice, hepatomegaly, failure to



thrive, lethargy, and bleeding diathesis. If not treated promptly, sepsis, liver failure, cataracts, intellectual disabilities, growth delay and death may occur. However, chronic or secondary complications are probable in older children and adults even with early and adequate therapy, including delayed growth, poor intellectual functioning, speech defects, motor problems, learning disabilities and ovarian failure.

To prevent the primary manifestations of galactosemia, it is very important to perform newborn screening tests for all infants and immediate restrictions on all lactose-containing foods and medicines in those affected. Symptoms resolve quickly and prognosis is good if dietary therapy is started in the first three to ten days of life. Routine monitoring of galactose accumulation is necessary to make adjustments to treatment. Other interventions include calcium supplements, ophthalmologic examination, developmental evaluation, and speech assessment. For families at risk of having an affected child, genetic counseling and prenatal diagnosis are recommended.

### Fetal alcohol syndrome

Fetal alcohol syndrome, the most severe form of fetal alcohol spectrum disorders, is a preventable cause of intellectual disability. Fetal alcohol syndrome is the result of high alcohol consumption during pregnancy, especially in the first three months of gestation, which can inflict considerable harm to the developing fetus, particularly to the brain. Rates vary according to alcohol consumption in a population; in the USA, it is estimated at 0.2 to 1.5 per 1000 live births have fetal alcohol syndrome (Centers for Disease Control and Prevention, 2009).

Medical practitioners' knowledge about this condition is low and detection is very poor, with many sufferers going undiagnosed. Together with a history of maternal alcohol intake, currently physicians still rely heavily on three clinical characteristics for diagnosis (Centers for Disease Control and Prevention, 2009):

- Facial abnormalities
- Central nervous system abnormalities
- Growth deficits.



Click on the picture to view a one-hour educational video on fetal alcohol syndrome.

More effective diagnostic tools for early detection using biomarkers, such as fatty acid ethyl esters in the meconium (Bearer et al, 2005), are under investigation.

Clinical symptoms vary depending on the amount, frequency and timing of alcohol exposure, maternal and genetic influences. Infants with fetal alcohol syndrome usually show growth retardation and a mixture of characteristic craniofacial anomalies, which are the hallmarks, including a flat philtrum, thin upper lip, short palpebral fissures, epicanthal folds, low nasal bridge, short upturned nose, ear malformations, and flattened maxilla. Central nervous system abnormalities are also commonly seen in fetal alcohol syndrome, which may include microcephaly, seizures, poor motor coordination, neurosensory hearing loss, cognitive and functional impairments. Furthermore, fetal alcohol syndrome may predispose to the development of mental health problems and substance use.

The only way to prevent fetal alcohol syndrome is to keep away from alcohol during pregnancy. Education for pregnant women or who are planning pregnancy is essential. In addition, screening questionnaires and biochemical markers for detection of maternal alcohol use are useful to detect risk behaviors and intervention to prevent further damage to the fetus. More research on this area is sorely needed (Ismail et al, 2010).

## DIAGNOSING INTELLECTUAL DISABILITY

According to both the DSM and ICD, three basic criteria should be met for a diagnosis of intellectual disability (or mental retardation):

- Significantly sub average intellectual functioning (IQ of 70 or below)
- Concurrent deficits or impairments in adaptive functioning in at least two 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 is before age 18 years.

The subtypes and their characteristics have already been described earlier in the chapter. Diagnosis requires a full professional assessment of intelligence and adaptive behavior. Children with ID are usually brought to professional attention because of their behavior rather than their low intelligence. Moderate and severe

### Intellectual disability and DSM-5

The revision of the DSM classification is proposing to change the name of this group of disorders to "intellectual developmental disorder" and less reliance on IQ for diagnosis.

According to this proposal, a diagnosis of intellectual developmental disorder would require:

- Deficits in general mental abilities such as reasoning, problem-solving, planning, abstract thinking, judgment, academic learning and learning from experience with an IQ two or more standard deviations below the population mean for a person's age and cultural group (IQ below 70).
- These deficits significantly impair functioning by limiting participation and performance in one or more aspects of daily living, such as communication, social participation, functioning at school or at work, or personal independence at home or in community settings. These limitations result in the need for ongoing support at school, work, or independent life.
- Onset during the developmental period.

forms are identified early because developmental milestones are markedly delayed. Milder forms often become apparent during primary school as a result of academic difficulties, or even later, during adolescence.

A comprehensive assessment should include detailed questioning about:

- The medical history of parents and family: genetic conditions, infections during pregnancy, prenatal exposure to toxins, perinatal injury, prematurity, and metabolic disorders
- Development: language and motor skills, socialization, understanding and calculation
- Environment in which the child is raised: education, resources and the family environment

Physical examination, by the mental health clinician or pediatrician, focusing on symptoms associated with ID. For example, appearance of the face (e.g., the flat, broad face of Down syndrome) (Conor, 1999) or Brushfield spots (small white or grayish/brown spots on the periphery of the iris also common in Down syndrome) that may alert the clinician to the possibility of ID.

*IQ measurement* is mandatory in all cases in which ID is suspected. IQ should be measured using, if at all possible, widely accepted tests that have been standardized for the specific – or culturally similar – population (e.g., normative data obtained in a German population should not be used for Chinese children). Widely used tests include the *Wechsler Intelligence Scale for Children* and the *Stanford-Binet Intelligence Scales*.

It is also useful to evaluate adaptive behavior. To do that professionals compare the functional abilities of a child to other children of similar age and education. There are many adaptive behavior scales available, such as *Vineland Adaptive Behavior Scales* and *Adaptive Behavior Assessment System-II*, but an accurate assessment of children's adaptive behavior requires clinical judgment as well.

Laboratory investigations (e.g., genetic testing) are indicated in all patients with ID, seeking to ascertain its etiology, which may have significant implications for treatment, prognosis and prevention. The extent of these investigations will largely depend on parents' resources and availability of these tests in the specific country.

### **Differential diagnosis**

Another term “developmental delay” is a broader concept than ID. It indicates that children show difficulties in one or more areas of adaptive functioning; this does not necessarily imply that they have significantly impaired cognitive abilities. In other words, although individuals with ID also show developmental delays, not all developmentally delayed children are intellectually disabled.

Among the most common conditions that can be misdiagnosed as ID are specific developmental disorders and school underachievement. In the former, children may appear developmentally disabled because they perform poorly in one or several academic skills (e.g., reading, arithmetic) when in fact they have an average IQ. In the latter, poor school performance mimicking ID may be due to other factors such as depression or school non-attendance (see Chapter



C.3). Although rare, the possibility of severe environmental deprivation, which may present with similar symptoms to ID (e.g., in severely deprived and understimulated children in orphanages), should also be excluded. Many children with autism spectrum disorders also have ID, however, they show the specific symptoms of the autism spectrum disorder. In these cases both diagnoses should be made.

### **Borderline intellectual functioning**

The boundary between “normal” and “below average” IQ is not rigid, as highlighted by the fact that psychosocial impairment is also required for a diagnosis of ID. Individuals with an IQ in the range of 71-85 – *borderline intellectual functioning*, about 7% of the population – are not impaired enough to warrant a diagnosis of ID, nevertheless they face considerable difficulties due to their limited cognitive ability. They may be able to perform day to day activities and a simple job without assistance but are more vulnerable to stressful life events and more likely to develop a psychiatric disorder as a result.

## **MANAGEMENT**

In all cases of ID, the crux of treatment is early detection and early intervention. As no specific etiology can be found in up to 40% of cases and many known causes cannot be cured, in the majority of cases, the aim of treatment is not a "cure" but to minimize symptoms and disability through reducing risk (e.g., helping individuals to be safe at home or school), teaching life skills, improve life quality and support families and carers. Detailed goals and modalities of treatment for each individual will largely depend on the cause and severity of ID and comorbid conditions.

### **Etiological treatment**

If the cause of ID is detected in a newborn, usually through screening, etiological treatment should be administered (e.g., in diseases such as PKU, congenital hypothyroidism and galactosemia) as already described.

### **Challenging behaviors and behavior intervention**

*Challenging behaviors* is a term used to describe comportment that interferes with the daily life of individuals with ID and their carers, reduce their quality of life and survival. These represent a wide range of problems that includes, among others, aggression, self-injury (such as head banging or ingestion or inhalation of foreign bodies), destroying objects, non-compliance, idiosyncratic habits (e.g., restricted range of foods), and socially inappropriate behavior. These problems frequently result in carers seeking medical help and can easily overwhelm families' ability to cope with and care for these young people, often resulting in rejection or, in more severe cases, institutionalization.

As in non-ID individuals, challenging behaviors serve a function and they are maintained or reinforced if the person with ID is successful in altering their internal or external environment through their behavior – such as by gaining attention, avoiding duties or demands, achieving access to preferred activities or objects or control over their own life, sensory feedback (e.g. hand flapping, eye

poking), and reduction of arousal and anxiety. The causes of challenging behavior are complex and include:

- Medical
  - Unrecognized pain or discomfort
  - Side effects of medications
  - Substance abuse
  - Physical illnesses such as epilepsy
  - Behavioral phenotypes specific for a syndrome
- *Dual diagnosis.* As already highlighted, intellectually disabled young people have higher rates of psychiatric disorder. A comorbid psychiatric illness (often referred to as “dual diagnosis”) occurs in about half of individuals with ID, the more frequent being ADHD, depression, autism and conduct problems, but schizophrenia and bipolar disorder also occur at least as often as in the non-ID population. Psychiatric illnesses like schizophrenia, depression and obsessive compulsive disorder present with roughly the same features in those with mild ID as in other people. However, recognition can be difficult in youth with moderate and severe ID, who tend to display more disorganized, unpredictable and difficult to understand symptoms, compounded by inability to describe their experiences:
  - Depression may present as withdrawn behavior, irritability and aggression
  - Manic episodes may present as distinct periods in which the youth with ID absconds, becomes boisterous, irritable or disinhibited
  - Aggression without clear precipitants and associated with bizarre behaviors suggestive of hallucinations or suspiciousness could suggest schizophrenia.

The existence of a family history of psychiatric illness such as schizophrenia or mood disorders may raise the suspicion. Timely identification and treatment of these comorbid conditions reduce disability, family burden and improve quality of life. There are questionnaires available that can help clinicians in the assessment of comorbid psychiatric problems such as the Developmental Behaviour Checklist (DBC; Einfeld & Tonge, 2002) a 96-item inventory, and the Aberrant Behavior Checklist (ABC; Aman et al, 1985), a 58-item questionnaire.

- Environmental
  - Problems in the living and working environment (e.g., lack of stimulation, family conflict, bullying)
  - Life events (e.g., change of school, death or separation)
  - Communication issues (e.g., inability to communicate, carers not attuned to the young person’s needs, inappropriate management that reinforces challenging behavior)
  - Life stages (e.g., puberty)

- Behavior serves a function or purpose for the person. Challenging behaviors are maintained if the person is successful in altering their internal or external environment through their behavior.

### ***Management of challenging behaviors***

People with ID and challenging behaviors or dual diagnosis are one of the most disadvantaged groups in most countries, very often receiving substandard care. One of the key roles of mental health professionals dealing with people with ID is the management of challenging behaviors. This requires considerable time, experience and skill. The starting point is to ascertain whether there are treatable causes (as listed above) and to conduct a behavior analysis. The settings in which problems occur can be the home, school or vocational training place, respite care facility or institution. Behavior analysis includes:

- A detailed description of the challenging behavior
- When and where does it take place (time, place, activity, context)
- The sequence of events and interactions with others: possible triggers, how others respond, do these responses reinforce the behavior?
- Skill factors (or lack of) involved
- Potential needs met by the challenging behaviors.

Once this is understood, a behavior intervention plan can be designed to address the problem. The first goal will be to ensure the safety of the child with ID and of the family or carers. The second would be to extinguish the undesirable behavior. In most cases this will involve families, teachers or workers in institutions, taking into consideration the family's needs, strengths and weaknesses, and often supporting and empowering them. Educating, helping and engaging the support network is essential for success.

### **Medication**

Medication is often used (probably overused; Branford, 1994) in the treatment of people with ID. Because mental health professionals are often consulted in crisis situations, medication may be prescribed in an ad hoc basis to deal with the crisis, when medication should optimally be used after a comprehensive assessment and as part of an overall treatment plan in which behavioral management and involvement of the family are essential (see above). The particular issues of informed consent in this population should also be taken into account.

Smaller doses than those typical in non-ID patients should be used – people with ID generally have more medical problems and are more sensitive to the side effects of drugs. Therefore it is wise to start with a very low dose and gradually increase it according to response and side effects.

Use of medications in this group is largely based on clinical experience rather than trial data. In the absence of controlled trial data, antipsychotics, antidepressants, mood stabilizers and other psychotropic medications should be used for the same indications as for children without ID, with the precautions listed above. The growth in the use of second generation antipsychotics, particularly for the management of challenging behaviors, is of concern. While some trial data in children (Snyder et al, 2002) – not replicated in adults (Tyrer et al, 2008) – suggest



Click on the picture to watch a video about education programs in people with ID (Cantonese with English subtitles; 8:01).



Click on the picture to watch a video on the US "Individualized Education Program" (IEP) (2:38).

that they may be useful in the short term, there are growing concerns about their long-term use because of side effects (e.g., metabolic syndrome).

*Agents that reduce sexual desire* are of special relevance to adolescents with ID because they occasionally display inappropriate, difficult to control sexual behavior. This can cause alarm and may result in restrictions to their freedom. Although the use such medications (e.g., cyproterone, testosterone antagonists) pose significant ethical challenges, they are increasingly prescribed for this purpose (Reilly et al, 2000).

### Education

In general, children with ID are less efficient at learning than other children. As they grow up and master activities of daily living, they need to attend school like other children. In high income countries such as the US, every child with ID from age three through 21 have access to free public education through an individualized education program. In a developing country such as India, there has been noticeable progress in addressing the challenges posed by ID. However, there is still a long way to go in low income countries in terms of developing effective, accessible, and affordable interventions (Girimaji & Srinath, 2010).

Overall, attending school is essential for children with ID to learn not only academic skills but also self-discipline, social and practical skills for community living. Though they have more difficulty learning, experience and research has shown that by applying the right educational techniques, many are capable of acquiring the basic skills of reading, writing, and arithmetic.

### *Inclusion versus segregation*

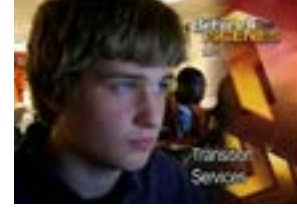
The current trend is to educate children with ID as far as possible in normal rather than special schools (inclusive education). This especially applies to those with milder forms of ID. However, there is limited evidence to compare the school experience of children with mild to moderate ID in mainstream and segregated education (Cooney, 2006). Another approach is to conduct special classes for children with ID in normal schools (opportunity classes). More severely retarded children may benefit from special schools. Whatever the approach, children with ID need education – even more so than other children – to maximize their development and chances in life.

### Physical and occupational therapy

Physical therapy and occupational therapy may contribute to treatment because ID is often accompanied by poor muscle tone, lack of coordination and slow development of motor skills. A therapist will be able to set up an individualized treatment plan that may be conducted at home, school or institution.

### Speech therapy

Speech and language are very important and highly specialized functions. They serve the crucial purpose of communicating one's own feelings and thoughts to others. Children with ID often show significant limitations in speech and language. Research has shown that a systematic application of speech therapy



Click on the picture to watch a video about children with disabilities' transition from school programs.



Click on the picture to watch a video about opening pathways to training and employment for people with intellectual disabilities in Africa



Click on the picture to watch a video clip from the the US Centre for Disease Control and Prevention about early detection of developmental disabilities (4:32).

techniques is effective in promoting speech, language and communication ability in ID children. Additionally, augmentative and alternative communication strategies can be used to help children with all levels of ID (Wilkinson & Hennig, 2007).

### **Family education and support**

Children with ID have the same basic human rights and fundamental freedoms as other children, including the rights to grow up in a family environment (United Nations, 2006). But the enhanced education and care needs of children can challenge the family's capacity to fulfill their functions and the ability to cope with exhaustion and isolation. Parents of children with ID generally reported needing more relevant information, psychological support and respite care services (Bailey et al, 1992). Support for families should meet these needs and prevent parents or other family members from feeling overwhelmed. To this effect, the WHO suggests that family support need to include communication of the diagnosis and information about it, emotional support, family counselling and training, involvement in health care decisions and respite care (World Health Organization, 2010).

## **PREVENTION**

Since ID is a life-long condition that cannot be "cured", prevention is very important. Prevention opportunities are summarize in Table C.1.3. Three levels of prevention can be distinguished: primary, which refers to a set of approaches that reduce or eliminate the risk of ID; secondary, which aims at an early diagnosis and treatment; and tertiary, which seeks to limit disability.

### **Primary Prevention**

#### *Genetic counseling*

Prospective parents, especially couples who already had a child with ID are usually keen to know the risk of future children being affected. Sensitive and accurate professional advice may help them make informed decisions about having other children. Such counseling can be as simple as telling parents who have a child with ID caused by a brain infection that the risk for their next child is very low, or it could be a very complicated matter needing costly tests when a genetic cause is suspected.

There have been rapid advances in the field of genetics. A set of techniques for the detection of genetic disorders called molecular genetics has evolved in the last decade. Though currently costly, the techniques are likely to become less expensive and applicable for wider use. One example is the possibility of detecting the presence of Down syndrome by performing a blood test on the mother during early pregnancy.

#### *Prenatal*

- Avoiding pregnancy before 21 years and after the age of 35 to reduce complications of pregnancy and labor. The risk of Down syndrome and other chromosomal disorders also increases after the age of 35. Prenatal screening/diagnosis of parents at risk.
- Abnormalities in the growing fetus can often be detected during early pregnancy. When a serious abnormality is found, parents may have



Click on the picture to watch a one-hour long video from the US Centre for Disease Control and Prevention about early detection of developmental disabilities.

the option of terminating the pregnancy. Some of these procedures are relatively safe, inexpensive and widely available (e.g., ultrasound). Others, such as genetic testing, are expensive, technically complex, not widely available and have their own risks.

### ***Perinatal***

- Pregnant women should add iodized salt to their diet to prevent iodine deficiency and avoid exposure to harmful chemicals and substances including alcohol, nicotine and cocaine.
- Neonatal screening. There are some causes of ID for which definite treatment is available in the form of medicines or special diets (e.g., PKU, galactosemia and hypothyroidism). Tests are available to detect these conditions at birth. If they are detected and treatment is started immediately, the occurrence of ID and other problems can be prevented.

### ***Postnatal***

- Universal immunization of children with the WHO schedule of recommended vaccines
- Prompt treatment for severe diarrhea and brain infections during childhood
- Providing a safe, caring, enriching and stimulating environment for children from infancy to ensure proper intellectual development.

### **Secondary prevention**

Some medical conditions associated with ID can be detected at birth. It is also possible to define a group of babies who are at risk of having a greater chance of developing ID as they grow up. These are children born prematurely, with a low birth weight (less than 2 kg), who have suffered asphyxia during birth, or have had a serious illness in the neonatal period. Following these infants up and monitoring whether they start to lag behind can be helpful in preventing ID.

By and large, most children with severe ID can be recognized by the age of 6-12 months. Mild ID usually becomes evident by the age of two years. Intelligence and social adaptation tests for the early detection of ID are now available, and can be adapted to any culture with appropriate modifications. Once a baby is suspected of or identified as having ID, it is necessary to provide appropriate stimulation for optimal development within that child's potential. These are techniques by which parents encourage and teach babies to use and develop their sensory (vision, hearing and touch) and motor (grasping, reaching, manipulating, and transferring) faculties. Such stimulation is necessary for normal development. Children with developmental delay need it all the more. Many manuals and guides have been developed for early stimulation, for instance, Portage Guide to Early Stimulation and Preschool Intervention for Developmentally Delayed Children (published by the National Institute for the Mentally Handicapped, Secunderabad, India). Some of these models have been successfully adapted (WHO, 2004).

### **Tertiary prevention**

#### ***Family***

The best place for children with ID to grow up in is with their own families. Organized support services are definitely needed for families to adapt well and face the situation with confidence and the least amount of stress. Parents need

#### **Associations and links**

- [American Association on Intellectual and Developmental Disabilities](#)
- [Australian Institute of Health and Welfare](#)
- [Australasian Society for Intellectual Disability](#)
- [Center for Effective Collaboration and Practice](#)
- [Council for Exceptional Children \(CEC\)](#)
- [Down's Syndrome Association \(UK\)](#)
- [European Association of Intellectual Disability Medicine](#)
- [Independent Living Canada](#)
- [National Center on Birth Defects and Developmental Disabilities \(US\)](#)
- [National Dissemination Center for Children with Disabilities \(US\)](#)

**Table C.1.3 Levels of prevention**

Level	Approach	Interventions
<b>Primary Prevention</b> (preventing the occurrence intellectual disability)	Health promotion	<ul style="list-style-type: none"> <li>• Health education, especially for adolescent girls</li> <li>• Improvement of nutritional status in the community</li> <li>• Optimum health care facilities</li> <li>• Improvements in pre, peri and postnatal care</li> </ul>
	Specific protection	<ul style="list-style-type: none"> <li>• Universal iodization of salt</li> <li>• Rubella immunization for women before pregnancy</li> <li>• Folic acid administration in early pregnancy</li> <li>• Genetic counseling</li> <li>• Prenatal screening for congenital malformation and genetic disorders</li> <li>• Detection and care for high-risk pregnancies</li> <li>• Prevention of damage because of Rh incompatibility</li> <li>• Universal immunization for children</li> </ul>
<b>Secondary Prevention</b> (halting disease progression)	Early diagnosis and treatment	<ul style="list-style-type: none"> <li>• Neonatal screening for treatable disorders</li> <li>• Intervention with “at risk” babies</li> <li>• Early detection and intervention of developmental delay</li> </ul>
<b>Tertiary Prevention</b> (preventing complications and maximizing functioning)	Disability limitation and rehabilitation	<ul style="list-style-type: none"> <li>• Stimulation, training and education, and vocational opportunities</li> <li>• Mainstreaming / integration</li> <li>• Support for families</li> <li>• Parental self-help groups</li> </ul>

to learn appropriate ways of rearing and training the child. They will continue to need assistance, guidance and support as the child grows up, especially during adolescence, early adulthood and during periods of crisis.

### **Community**

No program is likely to succeed without community involvement and participation. Services for individuals with ID include: medical and psychological (clinical) services. The first requirement is for appropriate facilities for a good medical/health evaluation and accurate diagnosis. Doctors should be in a position to recognize and manage treatable disorders such as hypothyroidism. Associated problems such as convulsions, sensory impairments and behavior problems can be corrected or controlled with proper medical attention. There are many claims that some drugs and herbal preparations can improve intelligence. But no drugs or any other treatment can cure ID. It is desirable to have facilities for psychological assessment of strengths and weaknesses in the child which can form the basis for future training. Adequate parental counseling in the initial stages is essential. Doctors, nurses, psychologists and social workers can make a big difference to parents by correctly explaining the condition, options for treatment, likely outcomes as well as by clarifying their doubts and helping them come to terms with having a handicapped child. Parental counseling also involves providing emotional support and guidance. Communities need to be educated also about the right of people with ID to lead their lives with respect and dignity, without discrimination. It is possible to achieve this goal by bringing about positive changes

in societal awareness, attitudes and beliefs about this condition. Individuals with ID should become an integral part of society, should not be isolated, segregated or discriminated against in any fashion.

Governments have the responsibility to provide optimum services to adequately address the needs of individuals with ID. This includes strengthening and effective utilization of existing services in the health, education and welfare sectors.

## PROGNOSIS

Many individuals with ID, particularly those of mild or moderate severity, are able to live independently and make a contribution to their community. Individuals with mild to moderate ID are frequently able to achieve some self-sufficiency and to lead happy and fulfilling lives. Individuals with severe ID are usually not able to live independently. Most people with Down syndrome who live into their 40s and 50s develop an Alzheimer's-like dementia (McPhee et al, 1999).

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