# Mental Retardation

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## DSM-IV Diagnostic Criteria for Mental Retardation

A. Significantly subaverage intellectual functioning: an IQ of approximately 70 or below on an individually administered IQ test (for infants, a clinical judgment of significantly subaverage intellectual functioning).

B. Concurrent deficits or impairments in present adaptive functioning (i.e., the person's effectiveness in meeting the standards expected for his or her age by his or her cultural group) 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.

C: The onset is before age 18 years.

*Code* based on degree of severity reflecting level of intellectual impairment:

**Mild mental retardation:** IQ level 50-55 to approximately 70 **.**

**Moderate retardation:** IQ level 35-40 to 50-55

**Severe mental retardation:** IQ level 20-25 to 35-40

**Profound mental retardation:** IQ level below 20 or 25

**Mental retardation, severity unspecified:** wheni there, is **a** strong presumption of mental retardation but the person's intelligence is untestabie by standard tests.

## ICD-10 Diagnostic Criteria for Mental Retardation

- Detailed clinical diagnostic criteria that can be used internationally for research cannot be specified for mental retardation in the same way as they can for most of the other disorders in Chapter V(F) of ICD-10. This is because manifestations of the two main components of mental retardation, namely low cognitive ability and diminished social competence, are profoundly affected by social and cultural influences. Only general guidance can be given here about the most appropriate methods of assessment to use. **Level of cognitive abilities**
- Depending upon the cultural norms and expectations of the individuals being studied, research workers must make their own judgments as to how best to estimate intelligence quotient (I.Q.) or mental age according to the bands given below:

| *Mental retardation* | *IQ range* | *Mental age (years)* |
|---|---|---|
| **Mild** | **50-69** | **9 to under 12** |
| **Moderate** | **35-49** | **6 to under 9** |
| **Severe** | **20-34** | **3 to under 6** |
| **Profound** | **Below 20** | **Less than 3** |

## ICD-10 Diagnostic Criteria for Mental Retardation

**Level of social competence**

Within most European and North American cultures, the Vineland Social Maturity Scale* is recommended for use, if it is judged to be appropriate. Modified versions or equivalent scales should be devel-oped for use In other cultures.

A fourth character may be used to specify the extent of associated impairment of behavior:

**No, or minimal, impairment of behavoir**

**Significant impairment of behavior requiring attention or treatment**

**Other impairments of behavior**

**Without mention of impairment of behavior *Comments***

A specially designed multiaxial system is required to do justice to the variety of personal, clinical, and social statements needed for the comprehensive assessment of the causes and consequences of mental retardation. One such system is now in preparation for this section of Chapter V(F) of ICD-10

## I.Q. Tests for Diagnosing Mental Retardation

| Intelligence Test | Age Range (years) |
|---|---|
| **Wechsler Preschool & Primary Scale of Intelligence-Revised** *(Wechsler, 1989)* | **3 to 7** |
| **Wechsler Intelligence Test for Children-lll** ***(Wechsler, 1991)*** | **6 to 17** |
| **Wechsler Adult Intelligence Scale-Revised** ***(Wechsler, 1981)*** | **16 to 74** |
| **Stanford-Binet Intelligence Scale: Fourth Edition** ***(Thorndike,*** *Hagen, and Sattler, 1986)* | **2 to adult** |
| **Kaufman Assessment Battery for Children** ***(Kaufman and*** *Kaufman, 1984)* | **2- 12** |
| **Kaufman Adolescent and Adult Intelligence Test** ***(Kaufman and*** *Kaufman, 1993)* | **11 to 85** |

## Common Measures of Adaptive Behavior

| Measure | Domains Tested |
|---|---|
| ***Vineland Adaptive Behavior Scales (Sparrow, Balla, and Cicchetti, 1984)*** | **Communication: receptive, expressive, written; daily living skills community socialization: interpersonal relations, coping skills; motor skills: fine, gross** |
| ***Scales of Independent Behavior-Revised (Bruininks, Woodcock, Weatherman, & Hill, 1996)*** | **Motor: fine, gross; social interaction and communication: language expression; personal living skills toileting, dressing; community, living skills money and value** |
| ***AAMR Adaptive Behavior Scales (Lambert, Nihira, and Leland, 1993)*** | **Independent functioning, physical development, economic activity, school** |
| ***Kaufman Functional Academic Skills (Kaufman and Kaufman, 1994)*** | **Functional math and reading skills, administered directly to person** |
| ***Street Survival Skills Questionnaire (Linkenhoker and McCarron, 1993)*** | **Administered to person; basic concepts, functional signs, tools, health** |

## Epidemiology

- Approximately 1 percent of the population has mental retardation. This 1 percent figure is cited by DSM-IV and is roughly the percentage found in most prevalence studies.
- Many have reasoned that mental retardation is more frequent-nearer to a 3 percent prevalence
- These controversies have enormous societal implications.

## Etiology I.

- **As noted by Esquirol in 1845, intellectual disability is not a disease in and of itself, but the developmental consequence of some pathogenic process.**
- **In 1898 William Ireland could classify idiocy into ten categories on the basis of etiology including "genetous, microcephalic, eclampsic, epileptic, hydrocephalic, paralytic, cretinism, traumatic, inflammatory, and idiocy by deprivation.**

## Etiology II.

- **With advances in medicine generally and in molecular genetics in particular, new causes of mental retardation or the genetic causes of formerly unspecified syndromes are identified each year.**

## Etiology III.

- **John Opitz counts over 750 genetic causes of intellectual disability alone.**
- **Eleanor Feldman notes that some 95 mental retardation syndromes have been linked to the X chromosome.**
- **The most common causes of mental retardation are Down syndrome, fragile X syndrome (ac-counting for 40% of all X-linked retardation), and fetal alcohol syndrome.**

## Mental Retardation Causes

| Cause | Examples | Estimated Frequency (%) |
|---|---|---|
| **Prenatal** | | |
| Genetic disorders | | 4-28 |
| Chromosomal aberrations | Down syndrome | |
| Single-gene mutations | Tuberous sclerosis, phenylketonu-ria fragile X syndrome | |
| Multifactorial | "Familial" mental retardation | |
| Malformation syndromes due to microdeletions | Prader-Willi, Williams, and Angelman syndromes | |
| Congenital malformations | | 7-17 |
| Malformations of the central nervous system | Neural tube defects | |
| Multiple malformation syndromes | Cornelia de Lange syndrome | |
| Exposure | | |
| Maternal infections | Congenital rubella, HIV | 5-13 |
| Teratogens | Fetal alcohol syndrome | |
| Toxemia, placental insufficiency | Prematurity | |
| Other | Radiation, trauma | |

## Mental Retardation Causes

| Cause | Examples | Estimated Frequency (%) |
|---|---|---|
| **Perinatal** | | |
| Infections | Meningitis | 2-10 |
| Delivery problems | Asphyxia | |
| Other | Hyperbilirubinemia | |

## Mental Retardation Causes

| Cause | Examples | Estimated Frequency (%) |
|---|---|---|
| **Postnatal** | | 3-12 |
| Infections | Encephalitis | |
| Toxins | Lead poisoning | |
| Other postnatal causes | Traumas, brain tumors | |
| Psychosocial problems | Poverty, psychotic illness | |
| **Unknown** | | 30-50 |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Down syndrome | Trisomy 21, 95% nondisjunction, approx. 4% translo-cation; 1/1000 live births: 1 :2500 in women less than 30 years old, 1:80 over 40 years old, 1:32 at 45 years old; possible overproduction of β-amyloid due to detect at 21 q 21.1. | Hypotonia, upward-slanted palpebral fissures, midface depression, flat wide nasal bridge, simian crease, short stature, increased incidence of thyroid abnormalities and congenital heart disease.<br>Passive, affable, hyperactivity in childhood, stubborn; verbal auditory processing, increased risk of depression, and dementia of the Alzheimer type in adulthood |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Fragile X syndrome | Inactivation of *FMR-1* gene at X q27.3 due to CCC base repeats, methylation; recessive; 1:1000 male births, 1:3000 female; Accounts for 10-12% of mental retardation in males | Long face, large ears, midface hypoplasia, high arched palate, short stature, macroorchidism, mitral valve prolapse, joint laxity, stra-bismus<br>Hyperactivity, inattention, anxiety, stereotypies, speech and language delays, I.Q. decline, gaze aversion, social avoidance, shyness, irrita-bility, learning disorder in some females; mild mental retardation in affected females, moderate to severe in males; verbal I.Q. > perfor-mance I.Q. |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Prader-Willi syndrome | Deletion in 15ql 2 (15q11 - 15q13) of paternal origin; some cases of maternal uniparental disomy; domi-nant 1/10,000 live births; 90% sporadic; candidate gene: Small nuclear ribonucleoprotein polypetide (SNRPN) | Hypotonia, failure to thrive in infancy, obesity, small hands and feet, microorchidism, cryptorchidism, short stature, almond-shaped eyes, fair hair and light skin, flat face, scoliosis, orthopaedic problems, prominent forehead and bitemporal narrowing<br>Compulsive behavior, hyperphagia, hoarding, impulsivity, borderline to moderate mental retardation, emotional lability, tantrums, excess daytime sleepiness, skin picking, anxiety, aggression |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Angelman syndrome | Deletion in 15q12(15ql 1 - 15q13) of maternal origin; dominant; frequent deletion of CABA B-3 receptor subunit, prevalence unknown but rare, estimated 1/20,000-1/30,000 | Fair hair and blue eyes (66%); dysmorphic faces including wide smiling mouth, thin upper lip, and pointed chin; epilepsy (90%) with char-acteristic EEG; ataxia; small head circumference, *25%* micro-cephalic<br>Happy disposition, paroxysmal laughter, hand flapping, clapping; pro-found mental retardation; sleep disturbance with nighttime waking; possible increased incidence of autistic features; anecdotal love of water and music |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Cornelia de Lange syndrome | Lack of pregnancy associated plasma protein A (PAPPA) linked to chromosome 9q33; similar phe-notype associated with trisomy 5p, ring chromo-some 3; rare (1/40,000-1/100,000 live births); pos-sible association with 3q26.3 | Continuous eyebrows, thin downturning upper lip, microcephaly, short stature, small hands and feet, small upturned nose, anteverted nos-trils, malformed upper limbs, failure to thrive<br>Self-injury, limited speech in severe cases, language delays, avoidance of being held, stereotypic movements, twirling, severe-to-profound mental retardation |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Williams syndrome | 1/20,000 births; hemizygous deletion that includes elastin locus chromosome 7q11-23; autosomal dominant | Short stature, unusual facial features including broad forehead, de-pressed nasal bridge, stellate pattern of the iris, widely spaced teeth, and full lips; elfinlike fades; renal and cardiovascular abnormalities; thyroid abnormalities; hypercalcemia Anxiety, hyperactivity, fears, outgoing, sociable, verbal skills > visual spatial skills |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Cri du chat syndrome | Partial deletion 5p; 1/50,000; region may be 5p15.2 | Round face with hypertelorism, epicanthal folds, slanting palpebral fissures, broad flat nose, low-set ears, micrognathia; prenatal growth retardation; respiratory and ear infections; congenital heart disease-gastrointestinal abnormalities Severe mental retardation infantile catlike cry, hyperactivity, stereotypies, self-injury |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Smith-Magenis syndrome | Incidence unknown, estimated 1/25,000 live births; complete or partial deletion of 17p11.2 | Broad face; flat midface; short, broad hands; small toes; horse deep voice. Severe mental retardation; hyperactivity; severe self-injury including hand biting, head banging, and pulling out finger and toe nails; ster-eotyped self-hugging; attention seeking; aggression; sleep disturb-ance (decreased REM) |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Rubinstein-Taybi syndrome | 1/250,000, approx. male ■ female; sporadic; likely autosomal dominant; documented microdeletions in some cases at 16p13.3 | Short stature and microcephaly, broad thumb and big toes, prominent nose, broad nasal bridge, hypertelorism, ptosis, frequent fractures, feeding difficulties in infancy, congenital heart disease, EEC abnor-malities, seizures Poor concentration, distractable, expressive language difficulties, per-formance I.Q. > verbal I.Q.; anectodally happy, loving, sociable, responsive to music, self-stimulating behavior; older patients have mood lability and temper tantrums |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Tuberous sclerosis complex 1 and 2 | Benign tumors (hamartomas) and malformations (ha-martias) of CNS, skin, kidney, heart; dominant; 1/10,000 births; 50% TSC 1, 9q34; 50% TSC 2, 16p13 | Epilepsy, autism, hyperactivity, impulsivity, aggression; spectrum of mental retardation from none (30%) to profound; self-injurious be-haviors, sleep disturbances |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Neurofibromatosis type 1 (NF1) | 1/2,500-1/4,000; male = female; autosomal domi-nant; 50% new mutations; more than 90% paternal NF1 allele mutated; *NFIgene* 1 7q11.2; gene prod-uct is neurofromin thought to be tumor suppressor gene | Variable manifestations; cafe au lait spots, cutaneous neurofibromas, Lisch nodules; short stature and macrocephaly in 30-45% Half with speech and language difficulties; 10% with moderate-to-pro-found mental retardation verbal I.Q. > performance I.Q.; dis-tractable, impulsive, hyperactive, anxious; possibly associated with increased incidence of mood and anxiety disorders |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Lesch-Nyhan syndrome | Defect in hypoxanthine guanine phosphoribosyl-transferase with accumulation of uric acid; Xq26-27; recessive; rare (1/10,000-1/38,000) | Ataxia, chorea, kidney failure, gout Often severe self-biting behavior; aggression; anxiety; mild-to-moder-ate mental retardation |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Galactosemia | Defect in galactose-1-phosphate uridyltransferase or galactokinaseorempirama se; autosomal recessive; 1/62,000 births in the U.S. | Vomiting in early infancy, jaundice, hepatosplenomegaly; later cata-racts, weight loss, food refusal, increased intracranial pressure and increased risk for sepsis, ovarian failure, failure to thrive, renal tubular damage Possible mental retardation even with treatment, visuo-spatial deficits, language disorders, reports of increased behavioral problems, anxi-ety, social withdrawal and shyness. |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Phenylketonuria | Defect in phenylalanine hydroxylase (PAH) or cofac-tor(biopertin) with accumulation of phenylalanine; approximately 1/11,500 births; varies with geo-graphical location; gene for PAH, 12q22-24.1; au-tosomal recessive | Symptoms absent neonatally, later development of seizures (25% gen-eralized), fair skin, blue eyes, blond hair, rash Untreated: mild-to-profound mental retardation, language delay, de-structiveness, self-injury, hyperactivity |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Hurler syndrome | 1/100,000; deficiency in a-L-iduronidase activity; au-tosomal recessive | Early onset; short stature, hepatosplenomegaly; hirsutism, corneal clouding, death before age 10 years, dwarfism, coarse facial features, recurrent respiratory infections Moderate-to-severe mental retardation, anxious, fearful, rarely aggres-sive |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Hunter syndrome | 1/100,000, X-linked recessive; Iduronate sulfatase de-ficiency; X q28 | Normal infancy; symptom on age set 2-4 years; Typical coarse faces with flat nasal bridge, flaring nostrils; hearing loss, ataxia, hernia common; enlarged liver and spleen, joint stiffness, recurrent infec-tions, growth retardation, cardiovascular abnormality Hyperactivity, mental retardation by 2 years; speech delay; loss of speech at 8-10 years; restless, aggressive, inattentive, sleep abnor-malities; apathetic, sedentary with disease progression |

## Mental Retardation Syndromes and Behavioral Phenotypes

| Disorder | Pathophysiology | Clinical Features |
|---|---|---|
| Fetal alcohol syndrome | Maternal alcohol consumption (trimester III>II>I); 1/3000 live births in Western countries; 1/300 with fetal alcohol effects | Microcephaly, short stature, midface hypoplasia, short palpebral fis-sure, thin upper lip, retrognathia in infancy, micrognathia in adoles-cence, hypoplastic long or smooth philtrum Mild-to-moderate mental retardation, irritability, inattention, memory impairment |

## Selected Clinical Findings and Laboratory Abnormalities That Increase Suspicion for Underlying Metabolic Disorder

- Growth abnormality
- Recurrent, unexplained illness
- Seizures
- Ataxia
- Loss of psychomotor skills
- Hypotonia
- "Coarse" appearance
- Eye abnormalities (cataracts, ophthalmoplegia, corneal clouding, retinal
- abnormality)

## Selected Clinical Findings and Laboratory Abnormalities That Increase Suspicion for Underlying Metabolic Disorder

- Recurrent somnolence/coma
- Abnormal sexual differentiation
- Arachnodactyly
- Hepatosplenomegaly
- Metabolic/lactic acidosis
- Hyperuricemia
- Hyperammonemia
- Low cholesterol
- Structural hair abnormalities
- Unexplained deafness
- Bone abnormalities (dysostosis, occipital horns, punctate calcifications)
- Skin abnormalities (angiokeratoma, "orange-peel" skin, icthyosis)

## Possible Contributions to Increased Vulnerability to Mental Disorders in Persons With Mental Retardation

- Neuropathological process responsible for mental retardation may also cause or increase risk for mental illness
- Increased likelihood *oi* loss or separation, particularly in out-of-home placements
- Communication deficits may predispose to emotional or behavioral disturbance
- Vulnerability to exploitation or abuse by others Inadequate coping skills
- Family stress may be heightened by presence of child with developmental disability
- Risk of limited network of social relationships and repertoire of social skills
- Risk of reduced opportunities for development and exercise of recreational and occupational skills
- Adverse effect on self-esteem of disability, possible dysmorphology

## Clinical Evaluation of the Patient With Mental Retardation

**Clinical history**
- Prenatal and birth history

**Family pedigree (three generations)**
- Relatives with learning problems, psychiatric disorders, mental retar-dation, neurological, or degenerative disorders
- Family resemblance (reduced in aneuploidy)

**Physical examination**
- Assessment of minor physical anomalies
- Growth and physical development Head circumference compared with norms Growth trajectory (comparison with earlier measures)
- Description of facial features (micrognathia, hypertelorism, thin upper lip); use of photographs or video, to document minor morphological variants, gait
- Complete neurological examination
- Documentation of behavioral phenotype
- Wood's light, dermatoglyphic examinations as indicated

## Clinical Evaluation of the Patient With Mental Retardation

**Adjunct diagnostics**
- Audiologic, ophthalmological, psychometric assessments

**Diagnostic tests (for selective use as indicated)**
- Skeletal radiographs
- Metabolic studies for lysosomal, peroxisomal, and mitochondrial disorders
- Muscle biopsies
- DNA molecular studies
- Chromosome analysis, fluorescent in situ hybridization (FISH)
- Fragile X testing
- Organic and amino acid assays
- Imaging studies (MRI, CT)

## Comorbidity

- ADHD
- Impulse Control Disorders
- Oppositinal Disorder
- Anxiety Disorders
- Eating Disorders (Pica)
- Psychosis
- Mood Disorders

## Treatment

- Prevention
- Psychotherapy
- Pharmacotherapy (antidepressants, anticonvulsants, anxyolytics, antipsychotics, psychostimulants, opioid antagonists)
- Services and Supports
- Schools for Handicapped Children
- Adult Services (summer camps, sport activity, Special Olympics)

Thank you for the attention!