Mental Retardation:
Overview and Diagnosis

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Author Dislosure
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Note: This is part 1 of a 2-part article. Part 2 will appear in July 2006.

Objectives

After completing this article, readers should be able to:

1. Contrast the current criteria used by the American Association on Mental Retardation and the American Psychiatric Association *Diagnostic and Statistical Manual of Mental Disorders IV* (DSM-IV-TR) to establish a diagnosis of mental retardation.
2. Characterize the relationship between the age of presentation and the severity level of mental retardation.
3. Recognize the importance of obtaining a detailed family history (three generations) as part of the etiologic evaluation of mental retardation.
4. Know the mechanism of inheritance for Fragile X.
5. List age-appropriate instruments for the measurement of intelligence and adaptive skills.

Introduction

Mental retardation (MR) is one of the more common developmental disabilities. It can be idiopathic and challenging to recognize in normal-appearing children who have developmental delays. Conversely, MR can be easily recognized when the child presents with dysmorphic features associated with a known genetic MR disorder. Mental retardation currently is defined by the American Association on Mental Retardation (AAMR) as "significantly sub-average general intellectual functioning accompanied by significant limitations in adaptive functioning in a least two of the following skills areas: communication, self-care, social skills, self-direction, academic skills, work, leisure, health and/or safety. These limitations manifest themselves before 18 years of age." (1) Recognizing that a numerical value alone may be neither precise nor adequate to distinguish between the abilities of a child whose intelligence quotient (IQ) is 71 and one whose IQ is 69, the AAMR defines the upper limit of subaverage general intellectual functioning as "70 to 75" when there are also significant concerns regarding adaptive abilities. The American Psychiatric Association *Diagnostic and Statistical Manual of Mental Disorders*, 4th edition–Text Revision (DSM-IV-TR) definition of mental retardation differs from that of the AAMR in that its cutoff IQ score for MR remains 70 (Fig. 1).

In addition to the controversy regarding the definition of MR, controversies exist regarding terminology for and determination of levels of MR. The term "mental retardation" is seen by some as stigmatizing and pejorative, although those who object generally concede that any new term would soon have similar negative connotations. Other advocates fear that a change in terminology might affect program eligibility, legal status, and treatment within the criminal justice system. The term eventually may change to "cognitive-adaptive disorders," but after much debate, the term "mental retardation" was maintained in the newest edition of *Mental Retardation: Definition, Classification, and Systems of Supports*. (1) Levels of MR traditionally have been based on the number of standard deviations (SD=15 points) below the accepted statistical mean IQ of 100. However, the exact numbers may vary by plus or minus 5 points in consideration of standard measurement error and according to the scoring protocols for various testing instruments.

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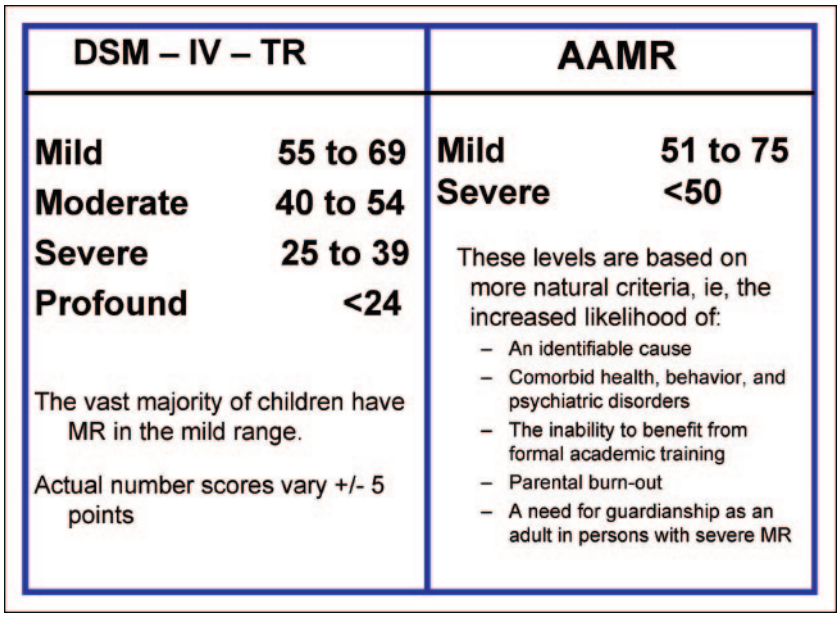

Figure 1. Levels of MR according to the DSM-IV-TR versus the AAMR.

Although the AAMR definition abandoned the traditional levels of MR classification (mild, moderate, severe, profound) in 1992, they were retained in the 1994 DSM-IV-TR definition. The 1992, the AAMR definition introduced a new system, "Intensity of Needed Levels of Support," to classify the severity of MR. This system was reaffirmed and expanded in the AAMR's 2002 definition. (1) The emphasis continues to be on the child's capabilities rather than on his or her limitations. The AAMR system is gaining support in the field, but it may not correlate well with IQ in some unique environmental situations. This categorization has been criticized by some for being less objective than the traditional classification systems for purposes of determining eligibility for services and for stratifying clients for research and legal purposes.

Aspects of the DSM-IV-TR stratification model (mild, moderate, severe, profound) still may be of value, particularly in defining the needs of individuals whose IQ scores are in the 50 to 75 range. For the purposes of this review, the term "mild MR" refers to individuals whose IQ scores are above 50, and the term "severe MR" refers to those whose IQ scores are below 50. This cutoff is clinically useful because it appears that individuals whose IQs are greater than 50 are more likely to benefit from a formal academic educational program; those whose IQs are less than 50 benefit more from an emphasis on life skills training.

Epidemiology

The statistical prevalence of MR is approximately 2% to 3%, although prevalence rates determined by ascertainment may be closer to 1% due to methodologic problems in identifying persons who have mild MR. The actual figures vary, depending on the study, ascertainment methods, age of cohort (lower prevalence in those <5 y and >18 y, ie, nonschool-age cohorts), and level of impairment. A stigma continues to be associated with the diagnosis of MR. Persons have an aversion to accepting it and tend to submit voluntarily to it only when a notable advantage is derived, such as cash support, school assistance, or additional services. The prevalence of severe MR remains stable at about 0.4% to 0.5%. The prevalence of mild MR, however, is more difficult to ascertain because the diagnostic limits are more variable. Nevertheless, most (about 85%) persons who have MR have IQ scores in the mild range. Many of such identified individuals "lose" their diagnosis of MR as adults, when demands on academic skills are fewer, they engage in vocational activities that rely on their adaptive strengths, and they blend in with and become indistinguishable from other members of the community.

The 2002 AAMR definition of MR has the potential to double the population of individuals identified as being mentally retarded because of its use of a range of scores rather than a distinct cutoff point (Fig. 2). Specific concerns have been voiced about the resulting impact of this "doubling" on eligibility determinations for special education services, Medicaid, supplemental Social Security Income, and other entitled programs. If this definition is used, 3% to 9% of individuals who have MR will require "extensive" or "pervasive" levels of support.

Causes

The number of known genetic causes of MR exceeds 1,000. This number is expected to increase as genetic techniques become more sophisticated and the ability to identify specific entities associated with cognitive impairment improves.

Although debate in the past surrounded the effects of "nature versus nurture" on cognitive development, it

now is clear that both play important roles. Researchers vary in their concepts of the dynamic interactions among genetics, the brain, and the environment, including experiences that result from the reactions of others to an individual's genetic phenotype. Adoption and twin studies have been used to investigate the impact of genes and environment, respectively.

Genetics appears to provide the cognitive potential that is shaped and developed by environmental and self-selected experiences that further modify one's behavior. At least 50% of the variance in IQ scores is gene-related. Heritability estimates for general intelligence appear to be about 0.45 to 0.75; longitudinal studies have shown this factor to increase steadily from infancy through adulthood. Different genes may play a role, each with a degree of effect (quantitative trait loci). The timing, intensity, and type of environmental experiences affect the underlying genetic potential. These factors may be biologic (eg, nutrition, lead, prenatal alcohol exposure, hypoxia) or social (eg, poverty, nutrition, degree of stimulation, maternal education). For example, low maternal education is the strongest predictor of mild MR; women who have fewer than 12 years of schooling are more likely to have a child who has an MR placement in school than are mothers who have some degree of postsecondary education. Women who have only high-school diplomas have an increased, albeit smaller, risk. Thus, genetic and environmental factors are interwoven and result in variable effects, depending on timing, intensity, and rate of experiences.

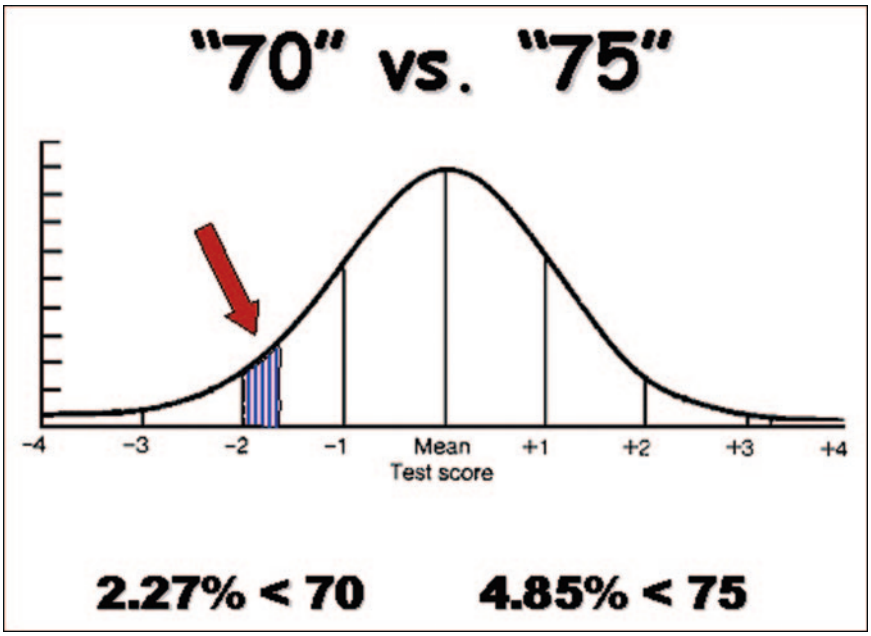

Figure 2. The additional number of persons identified as being mentally retarded when an IQ of 75 (vs IQ of 70) is used as the standard.

Numerous efforts have been undertaken to categorize MR based on the timing of the "cause": prenatal, perinatal, and postnatal. A limitation of exclusively using this temporal-based approach is that it requires the assignment of a single causative factor and does not consider the role of multiple events that may contribute to MR. Children who have birth defects are significantly more likely to be identified as having MR compared with children who have no birth defects, regardless of the type of defect. Such MR risks tend to be the highest among children who have central nervous system and heart defects.

The three most common identifiable causes for MR are fetal alcohol syndrome (FAS), Fragile X syndrome (FXS), and Down syndrome (DS). In individual children who have FAS, IQ measures range from 20 to 120, with a mean of 65. In comparison, IQ scores of affected male patients who have FXS range from 25 to 65, and those of children who have DS range from 40 to 60.

FAS is the most common preventable cause of MR. Children who have FAS exhibit deficits in measures of attention, learning, executive functioning, and visuospatial processing. Children affected with FAS who have normal IQ scores still may have significant neurobehavioral and adaptive deficits. Young adults who have FAS and normal IQs demonstrate deficits in the areas of attention, verbal learning, and executive function that are more severe than suggested by IQ alone. Their social abilities often plateau at the 6-year-old level, and their interpersonal relationship skills also are delayed. Compared with other individuals who have an identifiable cause for the MR, FAS-affected children are at an increased risk for behavioral and psychiatric disorders.

FXS is the leading cause of inherited MR, affecting approximately 50,000 persons in the United States alone (prevalence: 1 in 4,000 males; 1 in 6,000 females). FXS has been identified in every racial and ethnic group studied. Males who have the Fragile X full mutation usually exhibit moderate-to-severe intellectual impairment, characteristic language disorders (cluttering, receptive > expressive skills), and social and behavioral

difficulties, including problems with attention, impulsivity, anxiety, social avoidance, and arousal. There is, however, no relationship between the number of CGG repeats and IQ in males who have the full mutation. Affected males show a decline in cognitive, language, and adaptive skills measures during the school years. A specific cause for this decline is not known. Approximately 50% of females who have the full Fragile X mutation have MR. The remaining 50% may manifest borderline-to-normal intellectual functioning, learning disabilities related to executive functioning, or psychosocial difficulties. There does appear to be a relationship between IQ score and X activation scores or between IQ score and FMR1 protein levels in affected women.

The cognitive profile in FXS is similar in both affected males and females, with observed weaknesses in the areas of short-term memory for complex sequential information, visuospatial skills, planning, and verbal fluency. Many of these areas frequently are subsumed under the term "executive functioning." In FXS, the social deficits that comprise part of the full mutation phenotype range from autistic features to social anxiety and pragmatic language deficits. As many as 25% to 35% of individuals who have full mutations meet diagnostic criteria for autism.

The ability to identify children who have the Fragile X mutation by using genetic testing is of particular benefit to physicians and families. DNA studies for FXS should be strongly considered in every child in whom the cause of MR is unknown. The characteristic physical features of FXS are much more obvious in affected males, evolving over time such that they become more apparent during adolescence and adulthood. The two most frequently described findings are large ears and macro-orchidism. Other physical findings include a long and narrow face, a high-arched palate, loose connective tissue (hyperextensible fingers, flat feet), and mitral valve prolapse.

DS is the most common genetic disorder causing MR, occurring in 1 in 800 live births and in 1 in 1,000 conceptions. It usually is identified readily by characteristic physical features: hypotonia, hyperflexibility of joints, flat facial profile, slanted palpebral fissures, poor Moro reflex, excess skin on the back of the neck, abnormal ears, dysplasia of the midphalanx of the fifth finger, and single palmar crease. The diagnosis is confirmed by routine chromosome analysis, which reveals three possible abnormalities. The most common is a true trisomy for chromosome 21 (95%). Unbalanced Robertsonian translocations (3% to 4%) and mosaicism (1% to 2%) account for the other affected individuals. Most affected children have mild-to-moderate MR. Although the child who has DS continues to learn new skills, his or her measured IQ typically declines through the first 10 years after birth, reaching a plateau in adolescence that continues into adulthood. The profile of cognitive impairment in DS appears to differ from that of other forms of MR. Expressive language skills often are more delayed than cognitive and receptive language skills. Individuals who have DS also have relative impairments in their use of grammar. Affected individuals show relative strengths in visuomotor skills and relative weaknesses in auditory short-term memory.

Individuals who have DS have behavioral and psychiatric problems, but often less frequently than other groups of children who have MR. From childhood and into adolescence, the most frequent problems are disruptive behavior disorders, including attention-deficit/hyperactivity disorder (ADHD) and oppositional-defiant disorder. Approximately 7% also meet criteria for autism. As adults, individuals who have DS are more likely to have a major depressive disorder or demonstrate aggressive behaviors. A subset of older persons who have DS develops early signs of Alzheimer disease.

Clinical Presentation

The age at presentation usually is inversely proportional to the severity of MR. However, when dysmorphic features are present, especially those characteristic of a known genetic disorder, such as DS, MR may be suspected during infancy before developmental delays become apparent. Infants who have several minor congenital malformations have an increased likelihood of being diagnosed as being mentally retarded when they reach school age. Most children who have severe MR, regardless of appearance, are recognized within the first 2 postnatal years because they demonstrate obvious global delays in most developmental skill domains. With careful surveillance of language, visual problem solving, and adaptive skills, most children who have milder levels of MR can be detected by 3 to 4 years of age. Many children who have mild-to-borderline MR may elude recognition because their development during the early years approximates the lower limit of normal. These children may be diagnosed only after school entry, when they present with learning difficulties that prompt formal IQ testing.

Only rarely do parents raise a concern about MR to the pediatrician. Most affected children present with "speech" delay, the most common type of developmental delay. A busy pediatrician often sees several children who have "speech delay" each week. For the young child whose hearing is intact, language development is the best indicator of future cognitive abilities. In fact, one of the

most common causes of language delay is cognitive impairment or MR. When a parent is concerned about his or her child's speech, the pediatrician must determine whether the child has an isolated expressive language (speech) delay or a more serious combined expressive and receptive language delay. Due to time constraints and lack of pediatrician-friendly standardized tools, this determination is accomplished best by a speech and language pathologist.

The following delays (~ 30% or more) are definitely abnormal and always should prompt a referral by the physician for a thorough evaluation:

- Failure to turn to a voice by 6 months
- Failure to babble by 9 months
- Failure to orient to name at 13 months
- Failure to point to request or comment by 18 months
- Failure to follow a simple command without a gesture by 18 months
- Failure to use 10 to 25 single words by 24 months
- Failure to speak in two-word phrases by 26 months
- Failure to speak in three-word sentences by 36 months
- Unintelligible speech in a child older than 36 months
- Regression in language skills at any age

For more detailed developmental listings, the reader is referred to Johnson CP, Blascoe PA. Infant growth and development. *Pediatr Rev.* 1997;18:224–242.

All children who have language delays should undergo a formal audiologic evaluation, regardless of a normal newborn hearing screen. When both expressive and receptive skills are delayed, some measures of nonverbal intelligence and adaptive abilities are needed to differentiate MR from other causes of combined language delay. Hearing impairment and developmental language disorders are characterized by normal nonverbal and adaptive skills. Children who have autism demonstrate delays in social skills that are more severe than their overall level of functioning in other areas. Children who have delayed speech (expressive language) but normal receptive language do not have MR and have a good prognosis for "catching-up." Depending on the cause of their isolated expressive delays, such children may benefit from short-term speech therapy.

Sometimes, immature behavior prompts a visit to the pediatrician. Parents may become concerned when their child is unable to eat, dress, or toilet independently, especially when a younger sibling "passes up" the child or when these delays prevent admission into child care or preschool. During the early elementary school years, children whose mild MR remains undiagnosed may present with poor attention skills compared with classroom peers. Teachers may raise a concern about ADHD, although the child's ability to stay on task and concentrate actually may be consistent with his or her mental, rather than chronologic, age. Thus, the child may not meet criteria for ADHD, and medication treatment is not indicated.

Although children who have severe MR may have delayed motor milestones, motor development is not a reliable predictor of cognitive development. However, children who have primary motor delays are more likely to have neuromuscular disorders, such as cerebral palsy. Children who have mild-to-moderate MR usually master early gross motor milestones "on time." Subsequent, more complex gross motor skills and some fine motor skills may appear to be delayed due to the child's inability to comprehend verbal directions. Some children who have specific syndromes may demonstrate early gross motor delays due to associated abnormalities such as the hypotonia characteristic of DS. Stereotypic motor movements (similar to those seen in those who have autism) as well as self-injurious behaviors (head banging, self-biting) are more prevalent in children who have more severe levels of MR.

Diagnosis

The diagnosis of MR (or global developmental delay in younger children) involves two distinct and independent processes: 1) making a clinical diagnosis based on AAMR or DSM-IV-TR criteria and 2) searching for a specific medical cause. Although the clinical diagnosis of MR often requires the additional expertise of individuals beyond the traditional medical model sphere (psychologists, educators, therapists), efforts to identify the cause are coordinated best by the physician, usually with the assistance of a geneticist, neurologist, or a developmental pediatrician.

Establishing the clinical diagnosis of MR requires a standardized approach to determine whether key criteria are met, specifically, "significantly sub-average general intellectual functioning accompanied by significant limitations in adaptive functioning." This determination can be particularly difficult in children younger than 3 years of age because testing instruments used for this age group have not correlated well with later measures of IQ. Although IQ testing is possible during the preschool years (Table 1), the label "mental retardation" usually is not applied until the child reaches school age. At that point, formal IQ testing is considered more reliable and reflective of the child's long-term abilities. Prior to that

Table 1. Instruments Frequently Used to Measure Cognition

Bayley Scales of Infant Development III

- Provides a measure of behavior and development in very young children (1 to 42 months)
- Provides standard scores and age equivalents for individual developmental domains
- Is technically not a test of intelligence and does not predict future intelligence well

Wechsler Preschool and Primary Scale of Intelligence III

- Provides IQ scores for children 2.5 to 7.25 years of age

Stanford-Binet Intelligence Scale (5th Ed)

- Provides a composite IQ score for individuals 2 to 85 years of age

Kaufman Assessment Battery for Children II

- Provides a Mental Processing Composite IQ for children 3 to 19 years of age

Wechsler Intelligence Scale for Children (WISC-IV)

- Provides a Verbal, Performance, and Full-Scale IQ score for children 6 to 12 years of age

Table 2. Instruments Frequently Used to Measure Adaptive Skills

Vineland Adaptive Behavior Scales II (VBAS II)

- Assesses five major domains: communication, daily living skills, socialization, motor skills, maladaptive behavior
- For children birth to 19 years of age

Adaptive Behavior Scales II (ABAS II)

- Assesses all 10 specific adaptive skills areas specified in DSM-IV-TR and the 3 general areas specified by AAMR
- For persons from birth to 89 years of age

Scales of Independent Behavior-Revised (SIB-R)

- Comprehensive assessment of 14 areas of adaptive behavior and 8 areas of problem behavior
- For persons from birth to 80 years of age
- A version is available for use with persons who are visually impaired

time, a "diagnosis" of global developmental delay often is used, which usually is sufficient to access necessary and appropriate support services within the schools and through public agencies until 6, and in some cases 9, years of age.

The usefulness of adaptive measures (Table 2) is limited by the number of standardized items and the frequent need to depend on the history rather than on direct observation of what the child can and cannot do. The need to differentiate between "ability" and "opportunity" must be emphasized. Sometimes a child's relatively strong adaptive skills, especially for daily activities learned by rote, may cause the family to deny the diagnosis of MR.

Certain other conditions may affect the validity of standardized assessments. Recognized complicating factors include sociocultural test bias; the lack of testing materials or examiners in the child's native language; lower socioeconomic status; level of education, motivation, illness, or emotional factors at the time of testing; lack of rapport with tester; and associated motor, sensory, and communication deficits.

Simply defining "significantly sub-average general intellectual functioning accompanied by significant limitations in adaptive functioning" does not complete the diagnostic process. Recommendations from the AAMR, but not the DSM-IV-TR, emphasize the need for continuing evaluations to describe both the strengths and weaknesses of the individual who has MR across five specific dimensions: intellectual/adaptive, psychological/emotional, physical health, environmental, and social. Within each of these dimensions, the level of support needed to enable the individual to participate and to be included in community activities is established as either intermittent, limited, extensive, or pervasive.

Most cases of MR represent the effect of a static disorder. However, over time, children who have MR become more different from their peers because of their slower rate of cognitive and social development. This phenomenon must be differentiated from regression or loss of previously learned skills, which when present, always should stimulate a thorough investigation or re-investigation to rule out known degenerative disorders such as Rett syndrome or a storage disease.

The second component of the diagnostic process is the search for the cause of the MR. One of the key issues is determining the extent of an appropriate and thorough assessment. Efforts to identify a particular cause, despite many disorders having no specific therapeutic intervention, can be important for several reasons: 1) to focus the primary care physician's anticipatory guidance regarding common symptoms and complications, 2) to predict and manage associated conditions, 3) to determine long-range outcomes and prognosis, and 4) to provide appropriate counseling to the family regarding recurrence risk

in future pregnancies. Identification of a specific cause also may eliminate the need for additional unnecessary testing. Recently published consensus statements by professional organizations have attempted to address this somewhat controversial issue. (2)(3)

Despite the increasing number of diagnostic tests and procedures, the keystone to the search begins with the history and physical examination. It may be helpful to identify other family members who have learning problems, have had miscarriages or stillbirths, or who have consanguinity. A physical examination looking for dysmorphic features may suggest a cause (DS, cri du chat [5p-], velocardiofacial [22q deletion]). Just as there are unique physical phenotypes, there is an increasing body of knowledge about unique behavioral phenotypes that could suggest a specific syndrome (Williams, Smith-Magenis, Rett, Angelman).

A central focus of any diagnostic evaluation is determining whether a known genetic disorder is responsible for the individual's MR. A recent query of Online Mendelian Inheritance in Man listed more than 1,200 conditions that included MR as a clinical symptom and more than 250 molecularly identified MR genes. Single-gene disorders that cause MR may have an autosomal dominant (or arise by new mutation), autosomal recessive, X-linked recessive or dominant, and mitochondrial inheritance. Metabolic pathways, signaling pathways, and transcription are the most common functions controlled by these genes, but the genes also control numerous other aspects of neuronal and glial biology. Methods to examine genetic material have multiplied in recent years, although many still are unique to research laboratories. DS usually is detected with routine chromosome studies. High-resolution chromosome analysis (at least 500 G bands) may be necessary to detect more subtle chromosomal abnormalities (deletions, inversions, translocations, duplications) and should be a central component of the etiologic evaluation of every child whose MR is unexplained. Site-specific probing methods (fluorescent in situ hybridization) have been used when a particular syndrome or disorder is considered; each method has its own benefits and shortcomings. Recently, subtelomeric probes have received increasing attention. The subtelomeric region is a "gene-rich" area. These regions have the highest recombination rates and are prone to aberrant rearrangements during pairing and crossover between nonhomologous chromosomes. Rearrangements in these areas may result in specific clinical patterns in about 7% of individuals who have severe forms of MR. Due to the expense, this technique usually is tried only after negative results with routine chromosome testing have been obtained and with the consultation of a geneticist.

The appropriate use of imaging technologies is also controversial. Although neuroimaging has been recommended, a recent consensus statement (3) suggests that the absence of physical findings, such as microcephaly, macrocephaly, and focal motor abnormalities, in children who have MR decreases the likelihood of a positive result. In cases in which neuroimaging is performed, magnetic resonance imaging generally is the study of choice. Imaging studies also may provide information about the timing of an insult and help rule out specific disorders. Routine electroencephalography for children who have MR is not indicated unless clinical evidence suggests a seizure disorder.

Metabolic disorders cause MR in a small percentage of patients. However, an increasing number of disorders are amenable to treatment if identified at an early age. The number of newborn screening tests for metabolic disorders continues to expand, but the tests performed vary among the states, and what to include is controversial. Newborn screening results, if available, always should be reviewed as part of the evaluation of the child who has MR. Additional metabolic testing should be considered when history (parental consanguinity, family history of similar problems, developmental regression, episodic decompensation) or physical findings (hypotonia, declining growth, hepatosplenomegaly) suggest a specific cause. When focused screening techniques are used, the yield approaches 5% compared with approximately 1% when unselected metabolic screening is performed. However, in the absence of specific findings, routine screening for inborn errors of metabolism is not indicated in the initial evaluation of a child who has MR.

Conclusion

MR is a chronic condition that often has no readily identifiable cause or treatment. In nondysmorphic children, those who have more severe cognitive delays usually are identified clinically at earlier ages. Making a timely diagnosis of MR depends on a high degree of suspicion, especially in a child who looks normal and demonstrates only mild language and adaptive skill delays. Diagnosis is a two-part process that includes the clinical diagnosis of MR based on published criteria and a search for a cause. The pediatrician's approach to the diagnosis and evaluation of MR must take into consideration a variety of factors: genetic, environmental, and educational. Obtaining a detailed family history is a critical component of that process. Deviations by a child diagnosed as having MR from his or her expected devel-

opmental trajectory or a true regression in his or her abilities warrants reconsideration of the diagnosis and the previous evaluation. The introduction of new technologies and identification of new causes make the search a continual process.

National Resources and Web Sites

American Association on Mental Retardation (AAMR)
444 N. Capitol Street, Suite 846
Washington, DC 20001
Phone: (800) 424-3688 or (202) 387-1968
Fax: (202) 387-2193
Web site: www.aamr.org

The Arc of the United States
1010 Wayne Avenue, Suite 650
Silver Spring, MD 20910
Phone: (301) 565-3842
Fax: (301) 565-3843
Web site: www.thearc.org
E-mail: info@thearc.org

CDC National Center on Birth Defects and Developmental Disabilities
Web site: http://www.cdc.gov/ncbddd/

The National Association of Dually Diagnosed (NADD)
132 Fair Street
Kingston, NY 12401
Phone: (800) 331-5362 or (845) 331-4336
Fax: (845) 331-4569
Web site: thenadd.org
E-mail: info@thenadd.org

National Information Center for Children and Youth With Disabilities
PO Box 1492
Washington, DC 20013-1492
Phone: (800) 695-0285 (voice/TTY)
Fax: (202) 884-8441
Web site: www.nichcy.org
E-mail: nichcy@aed.org.

President's Committee for People with Intellectual Disabilities (PCPID)
The Aerospace Center
370 L'Enfant Promenade S.W.
Room 701
Washington, DC 20447
Fax: (301) 317-5897
Web site: www.acf.hhs.gov/programs/pcpid/index.html

Social Security Administration
6401 Security Boulevard
Baltimore, MD 21235
Voice hotline: (800) 772-1213
TTY Hotline: (800) 325-0778
Web site: www.ssa.gov

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PIR Quiz

Quiz also available online at www.pedsinreview.org.

1. The IQ level above which a child who has mental retardation (MR) is considered *most* likely to benefit from a formal academic educational program is:

   A. 30.
   B. 40.
   C. 50.
   D. 70.
   E. 80.

2. You are speaking to a group of junior medical students about children who are born with birth defects and have a much higher risk for MR. These MR risks are *highest* among those who have:

   A. Central nervous system and heart defects.
   B. Cleft palate and ear defects.
   C. Eye and renal defects.
   D. Gastrointestinal and skin defects.
   E. Liver and orthopedic defects.

3. Fragile X syndrome is recognized as the leading cause of inherited MR and should be considered in children in whom the MR cause is unknown. A physical finding that may further increase the suspicion of fragile X syndrome and the need to perform specific DNA studies for this condition is:

   A. Coloboma.
   B. Cryptorchism.
   C. Large ears.
   D. Micrognathia.
   E. Nail hypoplasia.

4. Children who have severe MR, even though they demonstrate no unusual physical features, *most* typically are recognized by age:

   A. 2 years.
   B. 3 years.
   C. 4 years.
   D. 5 years.
   E. 6 years.

5. A 30-month-old boy is referred to you by a family practice physician for concerns about his cognitive development. You tell the referring physician that the *best* indicator of the child's cognitive development is his:

   A. Age at becoming toilet trained.
   B. Age at walking.
   C. Fine motor skills.
   D. Language development.
   E. Visual skills.