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# Mental Retardation: Diagnosis, Management, and Family Support

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## Introduction and Clinical Diagnosis of Mental Retardation

**T**he direct and indirect economic costs associated with mental retardation in the United States are significant: 51.2 billion dollars (2003 dollars) for persons born in 2000.<sup>1</sup> This represents a significant and ongoing economic impact, especially since the beneficial effect of numerous intervention and prevention programs such as newborn screening for metabolic disorders (phenylketonuria, hypothyroidism) have likely already been realized by the time of this birth cohort. There continue to be multiple etiologies for mental retardation despite the successful interventions directed at these high-severity, low-frequency disorders. Lifetime indirect costs (ie, productivity losses) far exceed direct health and educational costs, emphasizing the need to address the physical, social, and economic factors that limit the functional participation of persons with mental retardation.

The number of children classified as “mentally retarded” receiving services in federally funded educational programs has stabilized over the past 9 years between 1.26 and 1.28%.<sup>2</sup> During this same period, the number of children identified as “learning disabled” or as having “autism” has increased. Educational support remains a primary intervention for individuals with all levels of mental retardation and is guaranteed under the Individuals with Disabilities Education Act.

It is for these reasons that better efforts to understand the reasons for mental retardation and to develop

effective strategies to both prevent, whenever possible, and minimize the impact of mental retardation on individuals, their families, and their communities continue to be important.

### *Theories of Intelligence*

The recognition that mental illness and mental retardation are different is a 20th century concept. The distinction was not previously clear; there was no significant effort to make such a distinction. Because of this indifference, individuals with very different disabilities were frequently treated inappropriately with little expectation for improvement or contributing to society.

Cognition, intelligence, and IQ are not synonymous. Associations between each of these terms often lead to confusion and disagreement when they are used interchangeably. Studies of individual differences in intelligence, primarily using standardized IQ measures, have accumulated evidence for a significant genetic influence: 50% of the IQ test scores variation can be attributed to genetic variation.<sup>3</sup> Experience and environment will, however, also influence this innate ability.

Intelligence is a combination of the ability to learn, to pose, and to solve problems.<sup>4</sup> The first efforts to assess intelligence in a standardized fashion did not occur until 1905 (Binet–Simon Scale). Learning, both informal and formal, can occur through any combination of experience, education, and training. With learning and its application comes the ability to resolve novel situations, to complete tasks and complex projects, and to be productive. Efforts to define intelligence are further complicated by differences in describing its key elements. Traditional psychological models of intelligence distinguish between verbal and nonverbal (perceptual organizational) skills, while neuropsychological assessment models include assess-

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ments of attention, auditory visual and tactile perceptual function, verbal and language functions, spatial/constructional processing abilities, memory and learning, and executive functioning.<sup>5</sup>

Some psychologists, such as Spearman and Cattell, viewed the construct of intelligence as a general capacity to acquire knowledge. They described that ability with either specific factors (“g” or “s”) or subtypes of intelligence (fluid, crystallized). In contrast, others, such as Guilford, Gardner, and Sternberg, viewed intelligence as a construct that included different types of distinct abilities present to different degrees in each person. Various schemas for these intelligence types have been proposed with some complementary and overlapping features.

### *Defining Mental Retardation*

There are two widely accepted formal definitions of mental retardation in the educational, medical, and psychological literature: the 2002 definition from the American Association on Mental Retardation (AAMR)<sup>6</sup> and the 1994 definition from the American Psychiatric Association’s Diagnostic Statistical Manual of Mental Disorders (DSM IV).<sup>7</sup> Each definition has its detractors and its proponents; they have, however, many more common features than differences. In each, the disability must originate before 18 years of age. Each also requires “significant subaverage performances” on standardized measures of intelligence and adaptive abilities. Interestingly, until 1973, a “significant subaverage performance” on measures of intelligence was 1 rather than 2 standard deviations below the mean.

There have been 10 definition revisions since the American Association on Mental Deficiency (AAMD, the predecessor of AAMR) first proposed a standard definition for mental retardation in 1908. Over time, the focus of these definitions changed from describing a state of global incompetence and incurability to a description of abilities as well as deficits in how affected persons function in a variety of everyday life contexts. This definition evolution paralleled an evolution in the terms used to describe individuals with mental retardation with less stigmatizing and less pejorative “labels.”

The AAMD first included the requirement for a standardized assessment of adaptive skills in 1959. Adaptive behaviors are the collection of conceptual, social, and practical skills that people have learned so they can better function in their everyday lives. Sig-

nificant limitations in adaptive behavior impact a person’s daily life and affect their ability to respond to a particular situation or to the environment.

The AAMR definition of 1992 incorporated several significant changes in its approach to “mental retardation” as a diagnostic entity. It abandoned the traditional “levels” of mental retardation based on standard deviations below the mean scores: mild, moderate, severe, and profound. These “traditional” levels, however, continued to be used in a majority of other settings and still remain an integral part of the DSM IV definition. AAMR adopted a two-tiered system to describe level of involvement based on the likelihood of an individual benefiting from formal educational services: mild and severe. The 1992 AAMR definition of mental retardation was also the first to use a range of intelligence scores (70 to 75) rather than a specific numerical score (70) to define the lower limit of “normal” when there were also significant concerns regarding adaptive abilities. The 1992 AAMR revision also introduced a new component to the definition, “Intensity of Needed Levels of Support.” A goal of this additional construct was to de-emphasize the impact that the numerical IQ score had increasingly assumed in future needs planning. This supports component evaluated the specific needs of the individual and suggested strategies, services, and supports that would optimize an individual’s functioning across four dimensions: intellectual abilities, adaptive behavior, participation, and health. The supports component recognized that individual needs and circumstances change over time. “Supports” can be technology, individuals, and agencies or service providers. A fifth support dimension, context considerations, was added in 2002.

The DSM IV (1994) definition of mental retardation continued to emphasize levels of severity based on standard deviations below the mean (mild, moderate, severe, and profound) and a single “cutoff score” (70) as central components of their definition of mental retardation. The reasoning has been that this classification schema provides a more reliable and reproducible framework for investigations and interventions around issues of organic etiology, risk factors, frequency of comorbid disorders, early intervention benefits, and adult outcomes. However, the more significant difference between the DSM IV and AAMR definitions may be the DSM’s retention of a specific cutoff score (70) rather than a scoring range (70 to 75) in the AAMR definition. Few would argue that there is

a significant difference in the abilities of an individual scoring 69 versus 71 on a standardized intelligence test. However, a shift in the absolute cutoff to define mental retardation from 70 to 75 would double the population who could potentially be determined “mentally retarded.” It is this kind of change that emphasizes the current, evolving approach to defining mental retardation and the need to look beyond “a number” as the primary characteristic of any future definitions.

### *Epidemiology of Mental Retardation*

The overall statistical prevalence of mental retardation is approximately 2 to 3%, although the actual prevalence may be closer to 1%. Historical efforts to characterize mental retardation have centered on defining a specific etiology (genetic, environmental, unknown), the timing of the event (pre-, peri-, post-natal, unknown), and/or the degree of involvement (mild, moderate, severe, or profound). As might be expected, different studies have produced variable results. The variances may be the result of differences in classification, study methodology, and/or application of the definition. The vast majority (approximately 85%) of persons with mental retardation have IQ scores in the “mild” range (DSM IV: 55 to 69; AAMR: 50 to 75). However, an exact prevalence of mild mental retardation (MR) is more difficult to ascertain, as the diagnostic limits and methods are variable. The category of “moderate” mental retardation (DSM IV: 40 to 54) does not exist in the 2002 AAMR definition, making estimates of current prevalence difficult. The prevalence of severe mental retardation (DSM IV: 25 to 39; AAMR: <50) is stable at ~0.4 to 0.5% and is more often associated with organic disorders, other medical conditions, and dysmorphic features.

### *Clinical Presentation*

The most common clinical presentation in children with mental retardation is delays in both receptive and expressive language. Mental retardation is not, however, the only reason for such delays. Other etiologies should be explored, as the type of appropriate interventions could be significantly different. Explanations such as bilingualism and a family history of “late talkers” should be diagnoses of exclusion and not an assumed explanation. In many cases, the child will not simply “grow out of it.” The breadth of potential causes of language delay makes some form of standardized screening by the primary care provider a

priority. In every case, the importance of medical providers listening to and addressing the concerns of a parent/care provider cannot be overemphasized.

Abnormal hearing should always be considered as a potential cause of language delay. Developmentally appropriate measures to assess hearing function should be utilized in every case; no child is “too young” to have their hearing tested. Careful observation should also be directed toward the other areas of development. Language delay in association with atypical social and play skills should always raise the possibility of the autism spectrum disorders as an etiology. Standardized measures to detect language differences are available, even for children as young as 18 months old.

For children with mild mental retardation, many will not be formally identified until they enter an academic setting where their difficulties with processing increasingly complex and novel information become apparent. This delay in diagnosis may be despite their earlier language or other associated delays. Moderate mental retardation in a child is often diagnosed between 3 and 4 years of age. The impact of their cognitive as well as social/adaptive differences is much more likely to bring them to the attention of parents and providers than their language delays alone. Severe mental retardation is often diagnosed by 1 year of age, especially when dysmorphic features are present. Additionally, the impact of their associated medical conditions and the greater likelihood of global developmental delays, including motor delay, may explain their earlier identification.

### *Diagnosing Mental Retardation*

Appropriate efforts to diagnose mental retardation should not simply define a specific etiology for the mental retardation. The most common known preventable cause of mental retardation is the fetal alcohol spectrum disorders with an incidence of 1 per 100 live births per year in the United States. The most common genetically inherited cause of mental retardation is Fragile X with a prevalence of 1:3600 in males; it is estimated that between 1:4000 and 1:6000 females have the full mutation. The most common chromosomal cause is Trisomy 21, occurring in 1 per 800 to 1000 live births. Focusing only on identifying a cause is a narrow approach that ignores other, more important aspects of an individual—defining what is required for that individual to be included in typical, age-appropriate activities in the home, school, and

community. The most appropriate method to establish a specific and comprehensive diagnosis of mental retardation utilizes the following three-step process.

**Standardized Measurement of Intelligence and Adaptive Skills.** Numerous instruments exist to accurately assess these areas. Tests should be chosen carefully, recognizing the adverse impact of cultural, language, and environmental factors and differences. Other considerations may include the impact of other associated physical disabilities on the accuracy of the testing: illness, hearing, vision, and motor abnormalities. The method chosen must match the purpose of the assessment. The motivation of the student and the skill of the examiner can also affect the validity of the results. Formal, standardized measures of intelligence do not correlate well with later assessments until approximately school age. Measures of adaptive skills often consist of questionnaires completed by a parent or care provider. These instruments have generally been standardized based on the timing of skill attainment in a normal population. Newer adaptive instruments are being developed that describe each individual's skills and are directly related to developing an appropriate support plan.

**Define the Individual's Strengths and Needs Across the Five Dimensions: Intellectual Abilities; Adaptive Behaviors; Social Roles, Participation, and Interventions; Health Concerns; Contextual Considerations.** Limitations must be considered in the context of an individual's community. That is, how would the child typically function in everyday life? What support is or is not available? An individual's limitations will coexist with strengths; both are essential factors in the supports equation. It is important to remember that the needs may change. Reassessments will be necessary as the identified variables change. How much support an individual requires will vary across these five dimensions, and support may be intermittent (as needed), limited (constant for a limited period of time), extensive (constant and life-long but not in all environments), or pervasive (constant, life-long, and in all environments).

**Develop a Plan to Provide the Supports and Services Considering Both the Level and Intensity of Need.** The plan is a natural outgrowth of identifying strengths and limitations. It is important to recognize that the level and types of supports will change over time. For individuals with mental retardation, defining an area of need and not doing anything to improve it is no more appropriate than implementing an imper-

sonal plan that is not based on a specific individual's needs. This effort is often incorporated into an Individualized Support Plan. It should identify available and appropriate medical, educational, community, and work resources.

### *Implications for Intervention*

New technologies have improved the ability of the medical community to better answer "why" an individual functions in the mentally retarded range. In some cases, answering this question has led to more effective treatments or interventions that limit the impact of a disorder. In others, answering this question allows professionals to better predict what an individual will or will not be able to do and to monitor and provide anticipatory guidance for expected complications or associated conditions that are seen with specific disorders. In others, the best that can be offered may be information regarding the risk of this condition recurring in other (current or future) family members.

More important than answering the "why" question may be society's answer to the "what" question. Given an individual's particular pattern of strengths and limitations, typical environment, and available supports, what can be done to insure that the child and family are included in typical activities and interactions as much as possible? While we may not be able to directly affect their disability, the goal will always be to limit the effect of the impairment (capacity to perform) by minimizing restrictions on activity and participation.

### **The "Search" for an Etiological Diagnosis**

Families need and deserve answers to the question "why did it happen?" Current evaluation strategies should allow for determination of an etiologic diagnosis in at least half of all children with mental retardation. Physicians have available an increasing array of testing modalities ranging from the well-known standard chromosome analysis to sophisticated genomic microarrays, targeted molecular testing, metabolic testing, and new cranial imaging techniques. Choosing appropriate, logical, cost-effective testing for an individual patient can be bewildering as the testing choices expand, and the myriad number of known genetic conditions for which testing is available continues to

increase. The availability of a clinical geneticist to aid in the evaluation is optimal but primary care providers, by circumstance or choice, often need or want to initiate such evaluations and can often achieve a specific diagnosis by targeted testing.

### *Why Ask "Why?"*

Multiple benefits for both the child and the family derive from determination of a specific etiology or diagnosis. For the family, knowing the diagnosis can relieve guilt. Family members often blame themselves, an undetected medical mishap at delivery, or benign environmental agents for their child's condition. In the absence of a specific diagnosis, reassurance that there is no basis for guilt or blame may be ineffective. At a minimum the family can anticipate an improved understanding of the time of onset and the likely pathogenesis of the retardation. Additionally, once the etiology is known, families can make informed reproductive decisions based on the inheritance pattern of the condition and explore possible options for prenatal diagnosis, if appropriate to their situation. This will also be helpful to adult siblings as they anticipate marriage and starting a family. A specific diagnosis allows the family to network with established support groups and connect with and learn from other families of children with the same or a similar disorder. Finally, knowing the etiological diagnosis allows the family to become experts on their child's disease or disorder and to become proactive in the medical and educational arenas.

For the child, the benefits of a known disorder include an understanding of the natural history of the condition and knowing when and how to conduct the appropriate monitoring for known potential complications. Knowing the diagnosis also prevents unnecessary testing, some of which might be invasive and/or expensive. Specific therapeutic, educational, and medical strategies that have been developed and individualized for various known disorders can be implemented. Finally, vocational planning is facilitated when a diagnosis with known adult outcomes is established.

### *Strategies and Protocols*

Two important consensus statements, one from the American College of Medical Genetics<sup>8</sup> and the other from the American Academy of Neurology,<sup>9</sup> address the utility of diverse diagnostic testing modalities. Not surprisingly, the consensus guidelines from medical

genetics stress family history, the dysmorphology examination, and targeted gene testing, whereas the guidelines from neurology emphasize cranial imaging and EEG interpretations as clues to the diagnosis. Both however are in substantial agreement as to the basic necessary evaluation of the retarded/developmentally delayed individual.

The yield of diagnostic testing varies widely in published studies, usually reported in the range of 40 to 60%.<sup>10-12</sup> In large part this variability is due to the specific study population, the age of the patients, and the year of the study. The causes of retardation in an institution obviously differ significantly from those established in an outpatient neurology or genetics practice. Another source of variability is the specificity of the diagnosis. Some authors might consider "hydrocephalus" to be a diagnosis, whereas others would not consider this a diagnosis unless confirmed by determination of its molecular basis, for example, an L1CAM mutation as the cause of X-linked hydrocephalus.<sup>13</sup> Most experts agree that the highest diagnostic yield is in the young child with anomalies or dysmorphic features. Family motivation and follow-through are also important as a diagnosis is often only achieved with long-term follow-up.

The etiologies include a broad range of more or less specific entities. These include chromosomal abnormalities, single gene defects, recognizable syndromes, teratogens, prematurity, perinatal and acquired conditions, and cultural familial retardation. As noted, chromosomal disorders continue to be the most frequent specific etiology and we can expect this category to grow as technology permits increasing detection of small chromosomal rearrangements, duplications, and deletions. Determination of the specific causes of autosomal-recessive and autosomal-dominant disorders will also increase due to utilization of molecular approaches, such as whole genome scans, to localize the genes of potential interest and focus efforts at gene identification. Recognition of the importance of genes on the X chromosome has led to identification of over 140 disorders associated with syndromic X-linked mental retardation, and in over half of these, causative genes have been localized. In nonsyndromic X-linked mental retardation, 24 X chromosome gene loci have been identified.<sup>14</sup>

### *History*

***Prenatal History.*** As in all areas of medicine, the search for the etiology of a child's retardation begins

with the history and physical. The prenatal history includes inquiry regarding potential teratogenic exposures such as alcohol, cigarettes, and street drugs. Prescription drugs of concern include isotretinoin, warfarin, and anticonvulsant drugs.<sup>15</sup> Maternal diseases such as myotonic dystrophy are important, if rare, causes of mental subnormality in offspring.<sup>16</sup> Mothers who have inherited a gene for adult onset myotonic dystrophy I with a repeat expansion size over 50 repeats, even when asymptomatic, are at 50% risk to transmit the gene. Myotonic dystrophy I is a cytosine-thymine-guanine (CTG) trinucleotide repeat disorder; the repeat size expands during maternal meiosis. If the infant inherits an abnormal allele that has expanded to >1000 CTG repeats, the infant will manifest congenital myotonic dystrophy characterized by hypotonia, respiratory problems, and later, developmental delay. Variable manifestations are seen with repeat sizes from 50 to 1000, with symptom severity correlating with repeat size.

The pregnancy history offers important clues to fetal well-being such as onset and strength of fetal activity, the amount of amniotic fluid, and the fetal presentation. Although breech presentation has many etiologies, it may be a sign of in utero hypotonia; all syndromes associated with hypotonia have an increased incidence of breech presentation. Neonatal adaptation is also important to assess. The infant with a prenatal problem in brain development from any cause may have depressed Apgar scores and evidence of early dysfunction such as poor feeding, hypoglycemia, hypothermia, seizures, etc. A careful history may reveal clues suggesting an in utero onset for these problems, making birth events less likely to be relevant or etiologically important.

**Presenting History and Review of Systems.** The history alone helps the clinician determine an etiological diagnosis in a small percentage of cases. Noting when the family first suspected a problem can be helpful, particularly in ruling out a progressive or neurodegenerative process. A child who was apparently normal until 2 to 3 years of age warrants quite a different approach than does one who has always been behind. A history of episodic illness may trigger evaluation for metabolic disorders, whereas a history of consistent but slow development can make this broad category of disease less likely. Discussion of the child's behavior can be critical in determining the diagnosis. Some features will be observed during the physical but specific recalled parental descriptions of

behaviors related to feeding, sleep, and personal interaction can be extraordinarily helpful. For example, abnormal early feeding behavior, excessive friendliness, and aversion to loud noises can suggest Williams syndrome, whereas a history of poor sleep, aggression toward others, and self-hugging may suggest Smith Magenis syndrome. Reviewing copies of therapy notes or school Individualized Education Plans (IEPs) can also be useful in documenting progress and developmental level. Parents may occasionally overestimate their child's accomplishments as part of a denial process. Parents are usually very concerned about behavioral issues and can relate these extensively and accurately. Instead of forging ahead to obtain a factual history as quickly as possible, it is helpful to focus these discussions on the most important areas and listen for the particular clues that may guide you toward a specific diagnosis.

The review of systems can also elucidate areas of concern not previously reported such as recurrent ear infections, intermittent high fevers, early teething, recurrent vomiting, or recalcitrant constipation. The typical skin picking seen in Prader Willi syndrome or the recurrent otitis seen frequently in Fragile X may be best elicited in the review of systems.

**Family History.** Family history also may provide clues as to causation of the child's problems. A history of prior pregnancy loss, either stillbirth or miscarriage, may be suggestive of a familial chromosomal imbalance, as would a history of other family members with mental subnormality. The possibility that the parents could be related should be explored. The discovery that both parents are from a geographically isolated region may suggest consanguinity even when no overt relationship is reported.

A history of retardation in mother's brother, uncle, or other maternally related relatives may suggest an X-linked condition. Psychological symptoms in male or female relatives such as anxiety, obsessive-compulsive disorder, and depression may suggest fruitful areas for investigation as would symptoms of progressive dementia. In recent years it has become clear that female carriers of X-linked conditions often exhibit subtle symptoms. Female carriers of Fragile X who have X chromosome CGG repeats in the range of 50 to 200 were previously thought to be relatively normal. We now know that these premutation carrier females may experience premature ovarian failure and psychological symptoms including excessive shyness, depression, and anxiety.<sup>17</sup> Males with the premutation in

Fragile X families were assumed to be completely unaffected but are now known to be at increased risk for a condition termed FXTAS (Fragile X Associated Tremor Ataxia Syndrome) characterized by Parkinson-like ataxia and progressive dementia after age 50.<sup>18</sup>

### *Physical Examination*

The physical examination differs from that of the typical pediatric examination in that attention is focused on assessment for dysmorphic features, the neurological examination, and the behavioral phenotype. Many observations leading to a diagnosis may be obtained by merely watching the child. This optimally should occur before the child is undressed or measured. For example, the hand-wringing behavior in Rett Syndrome may become obvious even when this has not been reported as a concern. Similarly, the truncal ataxia and frequent laughing characteristic of Angelman syndrome can be observed best when the child is comfortable.

Growth parameters should be plotted and serial growth charts evaluated. Measurements of inner canthal distance, interpupillary distance, palpebral fissure length, ear length, hand and middle finger length, and penile length are important in objectively assessing specific features. References for measurements are readily available.<sup>19</sup>

Pictures of the child should be obtained as a routine part of every assessment. Permission for photos can be obtained at the time of clinic registration. As a diagnostic aid they are often more valuable than any laboratory test. They are of great help in the longitudinal assessment as phenotypes often evolve over time. Subtle changes can sometimes be captured by reviewing family photos or those taken serially in clinic.

The neurological status of the young child can again best be assessed by observation of gait and tone as well as the child's interactions with others. Holding and playing with the child offer valuable clues not obtained in the routine examination. Increasingly, genetic syndromes are identified by their characteristic behavior, as much as by specific dysmorphic features. Most microdeletion and duplication syndromes (eg, VeloCardioFacial Syndrome, Angelman syndrome, Williams syndrome) as well as many single gene disorders (eg, Rett syndrome, Joubert syndrome, Fragile X) appear to have distinctive behaviors, only some of which have been well characterized.

### *Choosing the Right Tests*

**Chromosomes.** Chromosomal disorders are the most numerically frequent cause of mental retardation and identification of these abnormalities has improved dramatically with technological advances. A routine karyotype at or above the 500 band level is indicated on every child with retardation, unless the cause is absolutely clear (such as in neonatal stroke or Neurofibromatosis I). Sometimes chromosome analysis can be helpful in ruling out aneuploidy even when a well-established syndrome is suspected, especially if the presentation is atypical or more severe than usual. When evaluating a child whose initial chromosome studies were done five or more years previously, and in whom a chromosomal error is still suspected, consideration should be given to repeating the study at a higher resolution and/or counting a greater number of cells to rule out mosaicism. Pigmentary abnormalities should lead to consideration of skin biopsy and a fibroblast karyotype to rule out somatic mosaicism.

**Fragile X Testing.** Fragile X is the most common inherited form of mental retardation, affecting both males and females, and may be as common as 1/3000.<sup>20</sup> It is known to be caused by a trinucleotide repeat expansion (CGG) that occurs in the fragile X mental retardation gene (FMR1) at Xq27.3. As in myotonic dystrophy, when a mother carries a small expansion (55 to 200 repeats), it can expand to a full mutation (>200 repeats) in maternal meiosis. Testing should be ordered in both boys AND girls when the cause of retardation is not known. The threshold for ordering this testing should be very low, since the typical phenotype of a long face, large ears, hyperextensibility, and macro-orchidism is often absent in the young child and the impact of this diagnosis on the family is significant. Exceptions to this rule include testing children with multiple congenital anomalies and those with microcephaly in which the yield is very low. The molecular test supplants cytogenetic techniques, which may be unreliable.

**Subtelomere FISH (Fluorescent In Situ Hybridization).** Subtelomeric FISH analysis uses DNA-specific probes to evaluate the tips of chromosomes where conventional banding studies are often difficult to interpret. Using panels of multicolored probes, analysis of the subtelomeres can be useful especially when a chromosomal abnormality is high on the clinician's differential and other routine investigations have not yielded a diagnosis. Several reviews have suggested

that the yield of such studies is in the range of 5 to 7%.<sup>21,22</sup> This technique as well as whole chromosome painting, in which the entire chromosome is coated with a fluorescent probe specific to a certain chromosome, can help in determining which chromosomes are involved as well as the break points of chromosomal translocations or other rearrangements. Before ordering subtelomeric probes, the child should have a high-resolution karyotype. Experts have suggested that the highest yield for subtelomeric studies is in those with moderate to severe retardation, short stature, microcephaly, subtle but definite dysmorphic features, pre- and postnatal growth retardation, and a positive family history for mental retardation.<sup>23</sup>

**Single-Site FISH.** Many chromosomal disorders in blood, bone marrow, and solid tissue are now detected rather routinely with the use of locus-specific FISH. FISH involves the use of site- or gene-specific fluorescent DNA probes which “stick,” eg, hybridize, to the matching single-stranded sequence on the specified chromosome. When viewed under the fluorescent microscope, the cytogeneticist can determine if the probe is present on both chromosomes of interest. For example, in a child with Williams syndrome the ELN probe on 7q will be missing on one chromosome 7, indicative of a deletion in this region. FISH testing is now used for many contiguous gene deletion syndromes, the most common of which are Williams (7q11), Prader Willi (15q11), VeloCardioFacial (22q11), and Smith Magenis (17p11).

When ordering testing, it may be useful to speak to the cytogenetics laboratory to make sure that one is ordering the correct FISH test. FISH has many other uses beyond the scope of this limited discussion.

**Comparative Genomic Hybridization.** Comparative genomic hybridization (CGH) is a new technique which is becoming clinically available in some centers. In experienced hands it is likely to be most useful in detecting microduplications and microdeletions (small partial trisomies or monosomies) beyond the resolution of usual cytogenetic techniques.<sup>24</sup> The technique utilizes a computer to compare the amount of DNA in each chromosome against the chromosomal DNA in a reference sample. Suspected gains or losses of genetic material can then be confirmed with FISH probes in the localized region of interest. Selected centers now offer subtelomere-, centromeric-, and locus-specific CGH arrays, whereas others offer whole genome array CGH with probes at approximately 1-Mb (approximately the size of one chromosome

band) intervals. The costs may soon approximate that of subtelomere FISH and may partially supplant that technology. At present, problems with interpretation, background “noise,” expense, and availability should make this a test “of last resort,” ordered by a clinical geneticist.

**Metabolic Testing.** Expanded newborn screening is now available in 23 states in the United States and screens for 20 or more of the 29 conditions recommended by the American College of Medical Genetics. This has allowed for identification and treatment of inborn errors of metabolism in many children who might otherwise have died or become handicapped. Although 15 states screen for less than 10 conditions, screening for congenital hypothyroidism is universal as is screening for phenylketonuria. These widespread public health measures have decreased the chance that a developmentally delayed child has an undiagnosed metabolic error and have made unselected screening for metabolic disease unnecessary and unlikely to yield a diagnosis. Multiple studies assessing the yield of routine metabolic investigations in the child with mental retardation have concluded that there is little utility in such screening.<sup>8,9,25</sup> However, targeted screening for specific metabolic disorders not detected in newborn screening programs is extremely valuable in the setting of specific clinical signs and symptoms. Specifically lysosomal storage disorders, peroxisomal disorders, and glycosylation disorders are not detected in newborn screening programs. Symptoms that should prompt focused metabolic testing include hypotonia, failure to thrive, loss of milestones, ataxia, coma, seizures, recurrent unexplained illness in infancy, and occasionally, hearing loss. Clinical findings that should suggest the need for additional testing include hepatosplenomegaly, coarse features, hypoglycemia, metabolic and lactic acidosis, unexplained rashes, abnormal fat distribution, structural hair abnormalities, unusual odor, and specific skeletal X-ray changes.

Focused testing has allowed for the delineation of several new and important causes of mental retardation such as those caused by errors in creatine transport. Creatine is necessary for normal brain development and at least three errors of creatine metabolism have been shown to cause delay, autistic spectrum disorder, and seizures. Of these disorders one is X-linked, caused by mutations in SLC6A8<sup>26</sup> and two are autosomal recessive (AR). Screening for these disorders can be accomplished by measuring the urine

creatinine/creatinine ratio, and, if this is abnormal, determining levels of urinary guanidinoacetate methyltransferase (GAMT) and arginine:glycine amidotransferase (AMAT). Magnetic resonance imaging will also reveal this defect as the characteristic creatine peak will be absent.<sup>27</sup> Treatment of creatine deficiency syndromes with supplemental creatine has been somewhat successful in the amelioration of symptoms in the AR forms but not the X-linked form of the disorder.

**Cranial Imaging.** Cranial imaging continues to have an important supportive role in the diagnostic approach to a delayed child. Specific patterns of abnormal neuronal migration or abnormalities of the corpus callosum or posterior fossa may point the way to a diagnosis or date the onset of the problem or help confirm or rule out a suspected condition. In certain instances, as in Joubert syndrome and its variants, a sentinel abnormality, the “molar tooth sign”<sup>28</sup> may narrow the differential diagnosis. The yield of such testing increases in the presence of microcephaly, macrocephaly, seizures, or loss of milestones but can be occasionally helpful in normocephalic children. In general, given a choice of imaging modalities, magnetic resonance imaging (MRI) is superior to computerized tomography (CT). MRI is expensive and requires monitored sedation, which may be a deterrent. However, if abnormalities are seen on CT, they almost always need to be confirmed on MRI, and CT will not detect subtle migrational abnormalities. CT remains the imaging modality of choice in suspected congenital infection or craniostylosis.

**DNA Diagnostic Testing.** The availability of clinical molecular testing for inherited or de novo causes of mental retardation have increased exponentially and this trend will continue for the foreseeable future. Picking the appropriate test rests on the establishment of an accurate clinical diagnosis, which can often be facilitated by consultation with a clinical geneticist. DNA diagnostics remain, in general, very expensive and in many cases DNA testing may be unnecessary in the clinical setting. For many rare disorders, the molecular basis of the disorder may be known but DNA testing may be confined to a research setting. To ascertain the availability of clinical or research molecular testing, consult Gene Tests ([www.genetests.org](http://www.genetests.org)). For children with a defined and distinctive clinical phenotype (eg, Cornelia de Lange syndrome) molecular testing is adjunctive but not critical. Mutation analysis is most likely to be helpful when the child’s presentation suggests but does not confirm a definite

diagnosis, as is often the case in Rett syndrome, Angelman syndrome, and others that lack pathognomonic features. This testing also may be critical when a family wants carrier testing and/or prenatal diagnosis in a subsequent pregnancy. This testing is also most clinically useful when only one gene is responsible for the condition, eg, limited molecular heterogeneity. These guidelines for DNA testing are likely to change as clinical availability increases and costs decrease.

**Adjunctive Testing.** Evaluation of mental retardation almost always necessitates additional evaluations other than those discussed above, both for diagnostic assistance as well as for planning for therapy and management. Ophthalmology, neurology, and dermatology are frequently needed consultations. Audiology testing often is indicated. A wide variety of other testing modalities (muscle biopsy, psychological testing, nerve conduction, specific laboratory testing, EEG, etc.) may be necessary steps in the multidisciplinary evaluation of an individual child but are beyond the scope of this discussion.

### Summary Testing Recommendations

1. Every child with MR deserves a careful clinical evaluation and counseling to attempt to determine the etiology and date the onset of the problem and to establish and convey recurrence risks.
2. A cytogenetic study at or >500 bands is almost always indicated; consider repeating if over 5 years since study and a chromosome abnormality seems likely. Consider skin biopsy in the presence of pigmentary abnormalities.
3. Fragile X molecular testing should be performed in all children with undiagnosed MR except in those with microcephaly or multiple anomalies.
4. Targeted FISH studies are indicated when a specific microdeletion syndrome is suspected.
5. Subtelomeric FISH and array CGH should be considered in consultation with a clinical geneticist.
6. Cranial imaging, especially MRI, may be diagnostically helpful especially in the presence of cranial contour abnormalities, microcephaly, macrocephaly, and neurologic symptoms.
7. Molecular testing is increasingly useful in confirming diagnoses, especially when prenatal diagnosis is an option for the family.
8. Adjunctive testing may include hearing, vision, psychological testing, and other specialized testing as indicated by the child’s presentation. Achieve-

ment of a diagnosis is often complex, requiring a multidisciplinary effort over time.

Families should expect a through diagnostic evaluation of their delayed child. Technological advances promise that an accurate etiologic diagnosis will be possible in an increasing number of children.

## Management

Management of the child with MR, as with any chronic condition, should not only focus on the child and his condition, but also on the family. The family is the child's best resource. Supporting the family and ensuring its emotional and physical health is an extremely important aspect of overall management. Thus, "management" in this document is divided into two sections: child-centered strategies and family-centered strategies. This section will focus on the child; family-centered strategies are discussed in the next section.

The four major aspects of caring for children with MR include (1) health (growth, developmental, and behavioral surveillance, and mental and dental health); (2) developmental and educational interventions; (3) community integration through social and recreational activities; and (4) special considerations in adolescence and transition to adulthood. Management of children with MR varies depending on presence or absence of a known syndrome and on the severity of MR. As noted above, one of the advantages of the etiological diagnosis is the availability of a standard management "protocol" as has been developed for Down syndrome. These protocols are useful in guiding the clinician in surveillance and screening strategies for comorbid and secondary conditions.

Children with *mild* MR are more likely to have idiopathic MR and to be healthy. Thus, health care may vary little from typically developing children. Their degree of "diversity" may be somewhat more obvious in educational and community settings. For these children, transition to adulthood may require extra effort but they are usually able to live independently and may marry, have a family, and work in a competitive job. On the other hand, children with *severe* MR are more likely to have a known etiology complicated by characteristic medical, behavioral, and psychiatric comorbidities that challenge health management and may shorten life span. These individuals will usually require more intense special education as

well as additional supports to facilitate community integration and transition to adulthood. As adults, they are less likely to live independently, marry, and parent children.

Because children and adolescents with mild MR require few special services, much of the following discussion applies chiefly to those individuals with more severe levels of MR; these individuals represent the minority of persons with MR. Specific interventions relating to 10 common MR syndromes are listed in Table 1.

### Health Interventions

The first step in management of a child with MR is breaking the news in a sensitive, compassionate, and culturally appropriate manner. When breaking the news, it is important to emphasize the child's strengths as well as the deficits. It is also important to be realistic without taking away hope. If the child is less than 6 years of age at the time of diagnosis, and the child does not have a syndrome known to be associated with MR, it may be more appropriate to use the term, "global developmental delay." However, one problem with a physician using the term "developmental delay" in the case of true MR is the misconception that the "delay" is temporary and "catch-up" is implied. Thus, when counseling parents that the child will continue to progress albeit more slowly than their normal peers, it is important to also convey the message that this progress will continue until the "end" of the developmental years, which is considered to be about 16 to 18 years of age when the brain is fully myelinated. The term "mental retardation" should be reserved for later when standardized intelligence and adaptive testing can be done to confirm the diagnosis.

As with any child, children with MR will benefit from comprehensive health care within the context of a "medical home." The American Academy of Pediatrics (AAP) defines the "medical home" concept as health care that is accessible, continuous, comprehensive, family-centered, compassionate, and culturally effective.<sup>29</sup> In children with delays, it should also be developmentally appropriate. Parent-professional partnerships, built on a foundation of mutual responsibility and trust, are also important, especially in children with chronic disorders such as MR. The quality of these partnerships have been rated as one of the most important aspects of medical care by parents of children with chronic conditions.<sup>30</sup>

**TABLE 1.** Common causes of syndromic mental retardation

<b>Angelman Syndrome</b>	
Clinical Presentation	Severe global developmental delay by 6 months of age with gait ataxia and/or tremulousness of the limbs. Microcephaly (80%) and seizures (characteristic EEG) are common.
Prevalence	1/12,000 to 1/20,000
Range of MR	Majority of affected individuals function in the severe retardation range.
Diagnostic Test	<i>DNA and FISH:</i> Methylation for 15q11.2 and FISH. Deletion: 70–75%; Uniparental Disomy: 2–5%; Imprinting Defects: 2–5%; UBE3A mutations: 20–25%.
Comorbidities	Seizures (often before 3 years of age) Movement or balance disorder, usually ataxia of gait and/or tremulous movement of the limbs Unique behaviors/hyperactivity
Support	Angelman Syndrome Foundation (ASF); 3015 East New York Street, Suite A2265, Aurora, IL; 60504 Phone: (630) 978-4245, Toll-Free Phone: (800) 432-6435; Fax: (630) 978-7408; E-mail: info@angelman.org; Web site: <a href="http://www.angelman.org/angel/">http://www.angelman.org/angel/</a> <sup>31</sup> ASF is a national organization comprising families of those with AS, and the professionals who serve them. ASF increases awareness and advances the treatment of AS through education, information dissemination, and support for families and individuals. ASF conducts a biennial conference and funds research.
References	Buntinx IM, Clinical profile of Angelman Syndrome at different ages. <i>Am J Med Genet</i> 1995; 56:176–83. <sup>32</sup> Clayton-Smith J. Angelman syndrome: a review of the clinical and genetic aspects. <i>J Med Genet</i> 2003; 40(2):87–95. <sup>33</sup> Williams CA. Neurologic aspects of Angelman syndrome. <i>Brain Dev</i> 2005; 27(2):88–94. <sup>34</sup>
<b>Down Syndrome</b>	
Clinical Presentation	Hypotonia, congenital heart disease (40–50%), typical facial and limb features.
Prevalence	1/800 to 1/1000
Range of MR	Majority in the mild/moderate retardation range with mean IQ = 40–50
Diagnostic Test	<i>Routine chromosome analysis:</i> consider skin biopsy for determination of mosaics Trisomy 21 > 95%; Translocations and Mosaics <5%
Comorbidities	Eye or visual (60–75%); ear or hearing (50–60%); obesity (60–80%); constipation (44%); celiac disease (4–17%); dermatologic problems (50–60%); thyroid disease (30–40%); periodontal disease (adolescents ~90%); leukemia (~1%); pulmonary disease (especially infections and OSAS = 30–40%); seizures (10–15%); autism (7%); psychiatric (especially depression = 15–20%); atlanto-axial instability (15%)
Support	National Down Syndrome Society (NDSS); 666 Broadway, New York, NY 10012; Phone: (212) 460-9330, Toll-Free Phone: (800) 221-4602; Fax: (212) 979-2873; E-mail: info@ndss.org; Web site: <a href="http://www.ndss.org">www.ndss.org</a> <sup>35</sup> NDSS assists individuals with Down syndrome and their families through education, research, information referral, and advocacy. Services include a toll-free phone line, an annual conference, and training for professionals. NDSS publishes <i>Update</i> , a quarterly newsletter, and <i>UpBeat</i> , a magazine for teens and adults written by/for persons with DS. A web site is operated for teen and adults.
References	American Academy of Pediatrics: Health Supervision for children with DS. <i>Pediatrics</i> 2001; 107(2):442–9. <sup>36</sup> Leonard S. et al. Medical aspects of school aged children with DS. <i>DMCN</i> 1999; 41:683–88. <sup>37</sup> Roizen NJ. Medical care and monitoring for the adolescent with DS. <i>Adolescent Med.</i> 2002; 13:345–358. <sup>38</sup> Roizen NJ. The early interventionist and the medical problems of the child with DS. <i>IYC</i> 2003; 16:88–95. <sup>39</sup> Kent L, et al. Co-morbidity of ASD in children with DS. <i>DMCN</i> 1999; 41:153–58. <sup>40</sup>
<b>Fetal Alcohol Syndrome/Spectrum</b>	
Clinical Presentation	Growth deficiency (especially microcephally); dysmorphic facial features (especially smooth philtrum, thin upper lip and short palpebral fissures), and deficits in cognition, learning, attention, and/or memory. Confirmed maternal exposure required for full syndrome of FAS.
Prevalence	1/100 (all types)
Range of MR	Leading known preventable cause of MR
Diagnostic Test	<i>Clinical Diagnosis</i> by an experienced professional using one of three criteria sets
Comorbidities	Failure to thrive with or without feeding problems, ADHD, depression, Central Auditory Processing Disorder (CAPD), sleep disturbances, inappropriate sexual behavior, substance abuse problems, strabismus, congenital cardiac disorders, hearing loss, renal dysplasia
Support	National Organization on Fetal Alcohol Syndrome; 900 17th Street, N.W., Suite 910, Washington, D.C. 20006; Phone: (202) 785-4585, Toll-Free Phone: (800) 66NOFAS; Fax: (202) 466-6456; E-mail: info@nofas.org; Web Site: <a href="http://www.nofas.org/">www.nofas.org/</a> <sup>41</sup> NOFAS is committed to reducing the number of alcohol-exposed births through prevention, education, public awareness, education, advocacy, and intervention. NOFAS also provides information and referral services.

**TABLE 1.** Continued

<b>Fetal Alcohol Syndrome/Spectrum</b>	
References	<p>Hoyme HE. A practical clinical approach to diagnosis of fetal alcohol spectrum disorders. <i>Pediatrics</i> 2005; 115(1):39–47.<sup>42</sup></p> <p>National Institute on Alcohol Abuse and Alcoholism. Identification and Care Of FA-Exposed Children: A Guide For PCP's. NIH Publication #99-4369, 1999<sup>43</sup></p> <p>Kvigne VL, et al. Characteristics of children who have full or incomplete FAS. <i>J Pediatr</i> 2004; 145:635.<sup>44</sup></p> <p>Streissguth AP, et al. Risk factors for adverse life outcomes in FAS and FAE. <i>J Dev Behav Pediatr</i> 2004; 25:228–38.<sup>45</sup></p> <p>Steinhausen HC, et al. Behavioural phenotype in FAS and FAE. <i>DMCN</i> 2003; 45:179–82.<sup>46</sup></p>
<b>Fragile X Syndrome</b>	
Clinical Presentation	Males may have a characteristic appearance: large head, long face, prominent forehead and chin, large ears. Also have joint laxity and large testes (postpubertal).
Prevalence	<p>X-linked dominant</p> <p>16 to 25/100,000 males affected with the fragile X syndrome</p> <p>Prevalence of females affected with fragile X syndrome is presumed to be approximately one-half the male prevalence.</p>
Range of MR	Majority of affected males function in the moderate mental retardation, while affected females function in the mild mental retardation range.
Diagnostic Test	<i>DNA</i> : Detection of an alteration in the <i>FMR1</i> gene caused by an increased number of CGG trinucleotide repeats (>200 typically)
Comorbidities	<p>Behaviors often include tactile defensiveness, poor eye contact, perseverative speech, problems in impulse control, and distractibility.</p> <p>Possibility of mitral valve prolapse</p> <p>Ophthalmologic problems in 25–50% of cases</p>
Support	<p>The National Fragile X Foundation (NFXF); P.O. Box 190488, San Francisco, CA94119; Phone: (925) 938-9300, Toll-Free Phone: (800) 688-8765; Fax: (925) 938-9315; E-mail: NATLFX@FragileX.org; Web Site: <a href="http://www.nxf.org">www.nxf.org</a><sup>47</sup></p> <p>The NFXF and its affiliates promote awareness, advance research, conduct advocacy, and offer support to the Fragile X community. The NFXF conducts a biannual international conference and awards research grants.</p>
References	<p>Hagerman RJ. Fragile X syndrome: a model of gene-brain-behavior relationships. <i>Mol Genet Metab</i> 2001; 74(1–2):89–97.<sup>48</sup></p> <p>Willemson R. The fragile x syndrome: from molecular genetics to neurobiology. <i>Ment Retard Dev Disabil Rese Rev</i> 2004; 10(1): 60–7.<sup>49</sup></p> <p>Hagerman RJ. "Fragile X Syndrome" In:Management of Genetic Syndromes. Allanson J, Cassidy S, editors. John Wiley and Sons, 2005. p.251–65.<sup>50</sup></p>
<b>Noonan Syndrome</b>	
Clinical Presentation	<p>Early developmental milestones may be delayed.</p> <p>Short stature, Pectus excavatum, broad webbed neck, cryptorchidism.</p> <p>Cardiac anomalies (50%). Distinctive face with ptosis and hypertelorism, changing phenotype with age.</p> <p>Joint hyperextensibility and hypotonia.</p> <p>No particular syndrome of behavioral disability or psychopathology is observed.</p>
Prevalence	Autosomal dominant. 1/1000 to 1/2500
Range of MR	Up to one-third of affected individuals function in the mild mental retardation range.
Diagnostic Test	<p><i>Clinical and DNA</i>: 50% with missense mutation in PTPN11 gene (12q24)</p> <p>Diagnosis of NS is made on clinical grounds</p>
Comorbidities	<p>Congenital heart disease occurs in 50–80% of individuals</p> <p>Varied coagulation defects in a third of patients.</p> <p>Ocular abnormalities occur in up to 95% of individuals: strabismus, refraction errors, and ptosis are frequent</p> <p>Short stature—responsive, to growth hormone</p> <p>Hearing loss in one third—usually conductive.</p>
Support	<p>The Noonan Syndrome Support Group, Inc. (NSSG); P.O. Box 145, Upperco, MD 21155; Phone: (410) 374-5245, Toll-Free: (888) 686-2224 in the US; E-mail: <a href="mailto:info@noonansyndrome.org">info@noonansyndrome.org</a>; Web Site: <a href="http://www.noonansyndrome.org">www.noonansyndrome.org</a><sup>51</sup></p> <p>NSSG is dedicated to serving individuals touched by Noonan syndrome and the professionals who serve them. The organization disseminates information, provides opportunities for networking and support, promotes public awareness about NS, and supports research efforts.</p>
References	<p>Jongmans M. Genotypic and phenotypic characterization of Noonan Syndrome: new data and review of the literature. <i>Am J Med Genet A</i> 2005; 134(2):165–70.<sup>52</sup></p> <p>Zenker M. Genotype-phenotype correlations in Noonan syndrome. <i>J Pediatr</i> 2004; 144(3):368–74.<sup>53</sup></p>

**TABLE 1.** Continued

<b>Prader-Willi Syndrome</b>	
Clinical Presentation	Severe hypotonia, feeding difficulties in early infancy, excessive eating, and morbid obesity in later infancy or early childhood. Developmental delay. Short stature. Hypogonadism A distinctive behavioral phenotype is common: temper tantrums, stubborn, manipulative, and obsessive-compulsive characteristics.
Prevalence	1/12,000 to 1/15,000
Range of MR	Majority of affected individuals function in the mildly mentally retarded range (mean IQ: 60s to 70s), with approximately 40% having borderline retardation or low-normal intelligence and approximately 20% having moderate retardation.
Diagnostic Test	<i>DNA and FISH:</i> Methylation testing for the loss of paternally imprinted contribution in the 15q11.2-q13 (AS/PWS) region. Paternal deletion in 70%. Maternal Uniparental Disomy 28%. Imprinting Defect <2%.
Comorbidities	Morbid obesity unless it is externally controlled. Hypogonadism is present in both males and females Hypothalamic growth hormone deficiency → treat with GH → increase ht, decrease weight for height index and increase lean body mass, bone mineral density, and physical performance. Characteristic behavior profile becomes evident in early childhood in 70–90% of affected individuals: temper tantrums, stubbornness, controlling and manipulative behavior, obsessive-compulsive characteristics, and difficulty with change in routine.
Support	Prader-Willi Syndrome Association (USA); 5700 Midnight Pass Road, Suite 6, Sarasota, FL 34242; Phone: (941) 312-0400, Toll-Free Phone: (800) 926-4797; Fax: (941) 312-0142; E-mail: national@pwsausa.org.org; Web Site: <a href="http://www.pwsausa.org">www.pwsausa.org</a> <sup>54</sup> The PWSA (USA) and affiliates provide information, education and support services on behalf of persons with Prader-Willi syndrome, their families, and professionals who serve them. PWSA funds research grants and hosts an annual national conference.
References	Gunay-Aygun M. Changing Purpose of Prader-Willi syndrome clinical diagnostic criteria and proposed revised criteria. <i>Pediatrics</i> 2001; 108(5):E92. <sup>55</sup> Eiholzer U, Whitman BY. A comprehensive team approach to the management of patients with PWS. <i>J Pediatr Endo Metab</i> 2004; 17:1153–75. <sup>56</sup> Carrel AL. Growth hormone improves mobility and body composition in infants and toddlers with PWS. <i>J Pediatr</i> 2004; 145(6):744–9. <sup>57</sup> Cassidy S, McCandless S. Prader Willi Syndrome In: Management of Genetic Syndromes Allanson J, Cassidy S, editors. New York: John Wiley and Sons; 2005. p. 429–49. <sup>58</sup> Gene Reviews@ <a href="http://www.geneclinics.org">http://www.geneclinics.org</a> <sup>59</sup>
<b>Rett Syndrome</b>	
Clinical Presentation	Normal head circumference at birth with subsequent deceleration. Normal development during first 6–18 months of life. Rapid regression in language and motor skills. Hallmark is the loss of purposeful hand use and its replacement with repetitive stereotyped hand movements
Prevalence	X-linked dominant. 1/10,000 to 1/15,000. 95% of Rett syndrome cases are the result of de novo mutation. Mothers can be tested to rule out carrier status.
Range of MR	Majority of affected individuals function in the severe retardation range.
Diagnostic Test	<i>DNA:</i> MECP2 mutation analysis (Xq28) (positive in 85%)
Comorbidities	Feeding problems are common and poor growth is prevalent. Abnormal breathing and sleep regulation. Seizures are reported in 50%. Osteopenia and fractures in 33%.
Support	International Rett Syndrome Association (IRSA); 9121 Piscataway Road, Clinton, MD 20735; Phone: (301) 856-3334, Toll-Free Phone: (800) 818-RETT; Fax: (301) 856-3336; E-mail: <a href="mailto:irsa@rettsyndrome.org">irsa@rettsyndrome.org</a> ; Web Site: <a href="http://www.rettsyndrome.org">www.rettsyndrome.org</a> <sup>60</sup> An international organization, IRSA's members include parents of children with RS, caregivers, family members, and friends as well as physicians, therapists, and researchers. IRSA collects and disseminates information; lobbies for and funds research; conducts advocacy; offers support to families (print materials, conferences, help line, web site); and increases public awareness about RS.
Reference	Weaving LS. Rett syndrome: clinical review and genetic update. <i>J Med Genet</i> 2005; 42(1): 1–7. <sup>61</sup> Percy AK. Rett syndrome: clinical and molecular update. <i>Curr Opin Pediatr</i> 2004; 16(6):670–7. <sup>62</sup> Kerr A. Rett syndrome: recent progress and implications for research and clinical practice. <i>J Child Psychol Psychiatry</i> 2002; 45: 277. <sup>63</sup>
<b>Velocardiofacial Syndrome (22q11.2 Deletion)</b>	
Clinical Presentation	Typical craniofacial appearance, microcephaly, hypotonia, hearing abnormalities, and clefting anomalies/velopharyngeal insufficiency.

**TABLE 1.** Continued

<b>Velocardiofacial Syndrome (22q11.2 Deletion)</b>	
	Short stature. Conotruncal heart defects. Hypocalcemia common but resolves. Highly variable syndrome
Prevalence	1/2,000 to 1/6395
Range of MR	60% of affected individuals function in the mild to moderate MR range. There is a range of findings including learning difficulties (70–90%), especially of the nonverbal type.
Diagnostic Test	Full Scale IQ scores alone do not accurately represent the abilities of many of these individuals. <i>FISH</i> : Fluorescence in situ hybridization (FISH) using DNA probes from the DiGeorge critical region (DGCR) at 22q.11.
Comorbidities	Congenital heart disease (74% of individuals), especially conotruncal malformations. Palatal abnormalities (69%); velopharyngeal insufficiency in >90% with severe speech and articulation problems; immune deficiency (77%).
Support	Psychiatric and behavioral problems from depression to anxiety, OCD, and schizophrenia. Velo-Cardio-Facial Syndrome Educational Foundation, Inc.; 385 North Street, Weymouth, MA 02191; Phone: (781) 335-6652; Web site: <a href="http://www.vcfsef.org">www.vcfsef.org</a> <sup>64</sup> VCFSEF is an international nonprofit committed to offering support and information to individuals with Velo-Cardio-Facial Syndrome (VCF), their families and the professionals who serve them. VCFSEF educates the community about VCF, offers family support programs, encourages parent networking, publishes an informational newsletter, and hosts an annual conferences. VCFSEF also produces and disseminates educational materials.
Reference	Wang PP. Developmental presentation of 22q11.2 deletion (DiGeorge/velocardiofacial syndrome). <i>J Dev Behav Pediatr</i> 1998; 19:342–5. <sup>65</sup> The 22q11.2 deletion syndrome. <i>Adv Pediatrics</i> 2001; 48:39–73. <sup>66</sup> Robin RH. Defining the clinical spectrum of deletion 22q11.2. <i>J Pediatr</i> 2005; 147(1):90–6. <sup>67</sup>
<b>Williams Syndrome</b>	
Clinical Presentation	Pre- and postnatal growth retardation. Failure to thrive. Distinctive facial features and cardiovascular disease (elastin arteriopathy). Hypercalcemia and/or hypercalciuria are common. Characterized by cognitive impairment, unique personality characteristics (overfriendliness, excessive empathy, attention problems, and anxiety)
Prevalence	Autosomal dominant. 1/7500
Range of MR	Majority of affected individuals function in the mild mental retardation range.
Diagnostic Test	<i>FISH</i> : Detection of the contiguous gene deletion of the WS critical region (7q11.23), which can be detected using FISH.
Comorbidities	Supravalvular aortic stenosis (SVAS) seen in 75% of cases. This is the most clinically significant and most common cardiovascular finding. Idiopathic hypercalcemia (15%), hypercalciuria (30%), hypothyroidism (10%), early (but not precocious) puberty (50%), and failure to thrive in infancy (70%).
Support	Williams Syndrome Association (WSA); P.O. Box 297, Clawson, MI 48017-0297; Phone: (248) 244-2229, Toll-Free Phone: (800) 806-1871; Fax: (248) 244-2230; E-mail: <a href="mailto:info@williams-syndrome.org">info@williams-syndrome.org</a> ; Web Site: <a href="http://www.williams-syndrome.org/">www.williams-syndrome.org/</a> . <sup>68</sup> The WSA offers parent- and client-support programs; encourages and funds clinical research; engages in education and public awareness efforts; serves as an information clearinghouse; hosts a national convention; and conducts regional and local programs.
References	Mervis CB. Williams Syndrome: cognition, personality and adaptive behavior. <i>Ment Retard Dev Disabil Res Rev</i> 2000; 6(2):148–58. <sup>69</sup> American Academy of Pediatrics: Health care supervision for children with Williams syndrome. <i>Pediatrics</i> 2001; 107(5):1192–204. <sup>70</sup> Cassidy SB. Behavioral phenotypes in genetic syndromes: genetic clues to human behavior. <i>Advances in Pediatrics</i> . 49: 59–86, 2002. <sup>71</sup> Morris C. “Williams Syndrome” In: <i>Management of Genetic Syndromes</i> Allanson J, Cassidy S, editors. New York: John Wiley and Sons, 2005. p. 655–6. <sup>72</sup>
<b>Wolf-Hirschhorn Syndrome</b>	
Clinical Presentation	In more than 75% of affected individuals; distinctive facial features, cleft lip/cleft palate, IUGR/postnatal growth retardation, mental retardation, hypotonia, decreased muscle bulk, seizures, and/or distinctive EEG abnormalities and feeding difficulties
Prevalence	Previous estimates placed the prevalence of WHS at about 1/50,000 births, with a 2:1 female/male ratio. However, this appears to be an underestimation because of misdiagnosis and under recognition of affected individuals
Range of MR	Mild in 8%, moderate in 25%, severe in 67%.

TABLE 1. Continued

Wolf-Hirschhorn Syndrome	
Diagnostic Test	<i>Chromosome and FISH:</i> High-resolution G-banded cytogenetic studies detect the deletion in the distal portion of the short arm of one chromosome 4 involving band 4p16 in approximately 60–70% of individuals. FISH using a probe that includes the entire WHCR detects a deletion in more than 95% of individuals.
Comorbidities	The two most worrisome and difficult to manage medical problems in children with WHS are feeding difficulties and seizures. Severe failure to thrive and poor growth are common. G tube often necessary.
Support	4p16 Deletion Support Group (Wolf-Hirschhorn Syndrome); 1123 16th Avenue #51, Longview, WA 98632; Web site: <a href="http://www.4p-supportgroup.org">www.4p-supportgroup.org</a> <sup>73</sup> A nonprofit organization comprising families of children with WHS, the 4p16 deletion Support Group offers support to families, and publishes a quarterly newsletter. The group also coordinates a biannual national gathering and regional meetings. A listserv enables parents to communicate and share information with other parents of children with WHS and interested professionals.
References	Shannon NL. An Epidemiological study of Wolf-Hirschhorn syndrome: life expectancy and cause of mortality. <i>J Med Genet</i> 2001; 38:674–9. <sup>74</sup> Battaglia A. Health supervision and anticipatory guidance of individuals with Wolf-Hirschhorn syndrome. <i>Am J Med Genet</i> 1999; 89:111–5. <sup>75</sup> Battaglia A. Update on the clinical features and natural history of Wolf-Hirschhorn (WHS); experience with 48 cases. <i>Am J Hum Genet</i> 2000; 67(4):127. <sup>76</sup>

Comprehensive health care for any child, including the child with MR, should address growth, developmental and behavioral surveillance, anticipatory guidance and safety counseling, as well as traditional medical and dental care. In some children, psychiatric and therapeutic (physical, occupational, and speech therapies) services may be needed. Delivery of medical and dental care to a child with mild MR may be very similar to that of children of normal intelligence. Anticipatory and safety counseling should be modified to reflect the child's mental age rather than his chronological age

Providing care may be somewhat more challenging in children with severe levels of MR as they are more likely to be nonverbal and to have comorbid medical, behavioral, and psychiatric conditions. Physicians may be required to spend extra time and effort in communicating and coordinating care with subspecialists, school personnel, and community agency staff. Unfortunately, these extra challenges have resulted in decreased access to health care, and when it is available, it may be not only suboptimal but substandard. These stark disparities prompted a working conference of experts and a subsequent report entitled, *Closing the Gap: A National Blueprint to Improve Health of Persons with Mental Retardation*.<sup>77</sup> The "Blueprint" outlines goals to (1) improve community-based health care delivery for individuals with MR; (2) increase public knowledge and professional training regarding the unique health needs of individuals with MR; (3) improve health care financing; and (4) increase the quality and quantity of resources.

Finally, these conditions may present barriers to routine dental surveillance and preventative and restorative care, especially when the child is unable to cooperate and requires general anesthesia for simple procedures like cleaning and topical fluoride treatments. Recent emphasis on oral hygiene and dental care is evidenced by increased funding by Maternal Child Health for demonstration projects, the Surgeon General's report, and recent reviews on the topic.<sup>78</sup>

As noted above, children with more severe forms of MR, especially those with known syndromes, will more likely have specific comorbid disorders. Examples of disorders specifically associated with 10 common MR syndromes can be found in Table 1. Sometimes these disorders may impact their well-being and the provision of care to a greater degree than the cognitive impairment. Other comorbid disorders are generic and may occur in any child with MR. The following comorbidities may commonly occur in children with MR, especially in those with more severe degrees of MR.<sup>79</sup>

**Behavior Disorders.** The prevalence of behavioral problems increases as cognitive skills decrease. As many as 50% of those with severe MR will have significant behavioral challenges. Before labeling the behavior as "abnormal" or "deviant," it is first important to consider the child's mental age rather than the chronological age. For example, behavior representative of the "terrible twos" in a first grade boy with severe MR (eg, IQ = 35) is developmentally appropriate and expected, although it does, indeed, present a significant challenge to his teachers and caregivers.

When a new disruptive behavior occurs, one must also consider and rule out an organic etiology (eg, tooth abscess, stomach ulcer) especially in a child who is nonverbal and cannot localize pain. Behavior management strategies (at home and school) should be implemented. This is more challenging in the nonverbal child with more severe MR as these children are unable to communicate their frustrations or reasons for their challenging behavioral outbursts. When these strategies are not successful (and after the primary care physician has ruled out organic disease), often a referral to a developmental pediatrician and/or a child psychiatrist may be indicated for possible medication intervention. However, medical treatment should be just one component of a comprehensive management plan. Antiepileptic drugs (eg, valproate and carbamazepine) may be effective in treating irritability and mood swings. Some neuroleptics (especially newer generation ones with fewer side effects) may be helpful when aggression, self-injurious behavior, and stereotypies exist. When making decisions about the use of medications, one must also consider the negative impact they might have on rehabilitative interventions (for example, sedation while the individual is undergoing speech therapy).

**Psychiatric Disorders.** Comorbidity or “dual-diagnosis” is currently defined as a specific mental illness occurring in a person with MR. Mental illness is approximately four times higher in individuals with MR, especially severe MR, than in the general population. The more common comorbid psychiatric disorders include attention-deficit hyperactivity disorder (ADHD), anxiety, depression, obsessive compulsive disorder, oppositional defiant disorder, and, less commonly, schizophrenia. Diagnosing these conditions in children and adolescents with MR may be more challenging due to lack of reliable assessment tools and poor communication skills.

A comprehensive assessment should include the following<sup>80</sup>:

- A thorough history (health, family, developmental, behavioral)
- Concrete descriptions of behaviors, antecedent events, and environmental circumstances.
- A review of systems including personality patterns, adaptive functioning, communication, and social skills.
- Details of previous psychiatric interventions.
- A review of past cognitive evaluations.

- A patient interview that avoids the use of leading questions.
- A mental status assessment that is accomplished in the context of informal conversation rather than a formal examination—start with positives, ie, review interests and strengths and later focus on behavior problem (chief complaint).
- A review of medical records and assessment for the need for an etiologic evaluation especially when new diagnostic tests have been developed.
- Repeat cognitive evaluations when existing evaluations are not current and/or circumstances have changed significantly.

A DSM IV multi-axial diagnosis should be made if criteria are met.

Informed consent for psychotropic medications, especially those with significant side effects, is advised. When a medication is chosen, one should “start low” and “go slow.” Selective Serotonin Re-uptake Inhibitors (SSRIs) may be helpful for coexisting depression, anxiety, and/or obsessive compulsive disorder. Atypical neuroleptics may be used as a last resort in those with a psychiatric disorder. As with any patient, one should carefully monitor for side effects.

Attention deficits, impulsivity, and hyperactivity are common in school-age children including those with MR. Although the AAP Practice Guidelines for evaluation<sup>81</sup> and treatment<sup>82</sup> of ADHD do not address children with MR, it is reasonable to apply them to this population. Studies have shown that questionnaires and teacher input are valid<sup>83</sup> and reliable<sup>84</sup> in this population. One challenging diagnostic dilemma is determining the degree to which the child with MR is demonstrating true ADHD symptoms versus the degree to which a lower mental age is contributing to the deficits. When the child’s abilities to function in class appear to be due to ADHD, medical treatment may be indicated. Stimulant therapy has been found to be effective, especially in reducing hyperactivity,<sup>85</sup> although results may be less consistent in individuals with more severe forms of MR.

**Seizures.** Seizure disorders are approximately 10 times more common in children with MR, especially in those with severe impairments. Often the seizures are more challenging to control than those in typically developing children due to underlying syndromes, central nervous system pathology, and the coexistence of multiple seizure types in a single patient. If a child is prone to status epilepticus, an emergency protocol

should be established, which may include instruction in rectal administration of anticonvulsants at home and at school.

**Sensory Impairments.** Hearing and vision impairments are also more common in children with MR, especially in those with comorbid craniofacial syndromes. Approximately half of the children with severe MR will have visual deficits, the most common being strabismus and refractive errors.

**Motor Impairments.** Approximately 10% of those with mild MR and 20% of those with severe MR will have significant motor deficits consistent with cerebral palsy. On the other hand, approximately 50% of children with cerebral palsy will have comorbid MR.

**Sleep Disorders.** Sleep disorders are common in individuals with MR, especially those with severe MR and/or comorbid autism. Poor sleep is associated with poor daytime learning, poor occupational performance, and increased daytime behavior disorders, especially aggression and self-injurious behavior. Often, the most severe consequence of a sleep disturbance is the resulting physical and emotional burden on the child's family and caretakers. As with normal children, management should begin with behavioral strategies to improve sleep hygiene such as the implementation of consistent bedtime rituals, sleep scheduling, gradual distancing, extinction, and bedtime fading. Biologic interventions include melatonin and medications. Melatonin has been widely studied in children with disabilities and has been found to be very helpful in both inducing and prolonging sleep.<sup>86,87</sup> Melatonin is not yet FDA approved; thus, there is high variability among various brands. Short-acting preparations should be used when the child has difficulty falling asleep and long-acting preparations should be used for children with middle of the night awakenings. Some medications (eg, antihistamines, sedatives, clonidine) can be used on a long-term basis in children of all ages, while others (Ambien) are best reserved for older patients for short-term use during vacations, scout camp, and sleepovers with friends and/or relatives.

**Gastrointestinal Symptoms.** A variety of symptoms occur in children with all levels of MR; however, they are more difficult to diagnose and manage when the child has severe MR that is characterized by limited speech and ability to localize pain. Recurrent emesis may indicate a medical condition such as gastroesophageal reflux or, in rare cases, it may be the presenting feature of an underlying

metabolic disorder. In a nonverbal child, it may serve as a means to communicate GI pain or as an expression of anger, frustration, or anxiety. Thus, the first challenge is to discern whether the emesis is organic or behavioral in etiology. This may require the coordinated efforts of a gastroenterologist, a developmental pediatrician, a child psychologist, and/or a behavior specialist.

**Autism.** Autistic-like behaviors, for example, stereotypies, may be seen in some children with severe MR. Recently it has been shown that approximately 15 to 20% of children with known severe MR may also meet full criteria for autism.<sup>88</sup> Although not mentioned in practice guidelines published in the mid-1990s, it is now thought that about 7% of children with Down syndrome will meet criteria for autism.<sup>89</sup> In these children, social (in particular, joint attention skills) will be significantly more delayed than skills in other domains. Language skills are also more delayed than skills in other domains.

Children with a known etiological cause of MR may demonstrate any of the above comorbid disorders as well as additional medical conditions or a behavioral phenotype that may be unique to the syndrome. Some national, disorder-specific organizations (ie, National Down Syndrome Congress) have developed surveillance checklists to assist physicians and families to monitor for these conditions. When the child's symptoms are caused by a rare disorder that does not yet have a corresponding organization or web site, the National Organization of Rare Disorders (see Resource Guide) may be able to assist. Late secondary conditions associated with certain syndromes (ie, atlanto-axial subluxation in Down syndrome) may necessitate referral to a subspecialist or a multidisciplinary team of subspecialists. A few examples of these disorder-specific comorbid and secondary conditions for 10 syndromes can be found in [Table 1](#).

For the child with idiopathic MR, one may consider embarking on an additional etiologic search approximately every 5 years. New diagnostic tests and syndromes are described each year. Determining the answer to the "Why?" question can be comforting to parents and helpful in managing the older child for all the same reasons as those mentioned above for the original "search." However, clinical judgment guided by history and physical examination should be used so that tests are not obtained for no reason other than simply because they now are available.

## *Developmental and Educational Interventions*

In a healthy child with MR, the primary focus of overall management is almost always developmental and educational depending on the child's age. In either case, services should begin as soon as the delay or deficit is recognized. Families will need additional patience and persistence when raising a child with MR. Unlike their normal siblings, who seem to learn adaptive, language, manipulative, and play skills simply by modeling others, a child with MR may need deliberate and specific instruction to master a skill. When mastery is possible, it will occur at a much slower pace depending on cognitive level, behavior challenges, and comorbidities. Because of relatively concrete reasoning, a child with MR may have difficulty generalizing the new skill to similar tasks in different environments and situations. Finally, parenting style may affect learning; parents who overprotect their children impede their progress toward independence and negatively affect adult outcomes.<sup>90</sup>

**Early Developmental Intervention.** If a child is diagnosed with a developmental delay or a known disorder associated with delays or MR before the third birthday, he or she should be referred to a publicly funded EIP. These publicly funded programs are now "entitlement programs," so that any child with a delay or known disability has a right to services. An infant with dysmorphic features may be diagnosed with a recognizable syndrome (eg, Down syndrome) during the neonatal period. Even though developmental testing may not be effective in demonstrating significant delays during early infancy, MR can be assumed to be present if it is a major characteristic of the syndrome. Thus, the infant will become eligible for services at the time of diagnosis even though delays cannot be demonstrated.

On the other hand, a child without a recognizable syndrome will not be eligible until a significant delay (or delays) becomes evident. When significant delay is present, an etiological diagnosis is not necessary to access services; in fact, it is advisable to refer to an EIP as soon as possible, while the etiological evaluation is still in progress. Most children with MR have mild MR with subtle or unnoticeable delays and no physical abnormalities and are not usually diagnosed until after 3 years of age. Thus, most children with MR do not attend EIP programs.

On referral, the child will be scheduled for a multidisciplinary team evaluation that will confirm eligibil-

ity and serve as the foundation to develop an Individualized Family Service Plan (IFSP). The IFSP stresses the importance of the family as the central focus of and primary decision-maker for services affecting their child. The service menu may vary from state to state, but most programs offer case management, family support, parent training, and some direct therapy (speech, occupational, and physical). Ideally, services take place in a child's natural environment, ie, at home or in the child care center he or she attends.

Until 2005, children with global developmental delays or assumed MR associated with a known syndrome who were enrolled in an Early Intervention Program transitioned to a special preschool program on the third birthday. However, the new Individuals with Disability Education Act,<sup>91</sup> passed in December, 2004, allows the parents of these children to extend their child's IFSP to age 5 years instead of transitioning to the school system.

**Special Education.** Children with MR, especially mild to moderate MR without dysmorphic features and known etiological syndromes, are often not identified until after the third birthday. When this is the case, the clinician should refer the child to the "special education" department at the local school district. Occasionally, parents mistakenly contact regular preschool personnel who are not aware that delayed adaptive skills are characteristic of MR. If the child is not toilet trained, these school staff may tell parents that their child is not eligible for preschool services and admission is denied. This can be prevented if the pediatrician educates the parents about the differences between regular and special preschool programs and provides them with either a report with the developmental scores or a letter describing the pediatrician's concerns and the need for special services.

**Preschool.** Children 3 to 6 years of age are usually served in the "Preschool Program for Children with Disabilities (PPCD)." Services are free and usually half-day and bus transportation is provided. Unless the child has a known disorder diagnosed by a physician or significant developmental delays determined by standardized testing, a multidisciplinary team will evaluate the child to determine eligibility. The team's evaluation serves as the basis for development of an IEP, which takes place in a meeting of school personnel, especially those who participated in the evaluation, and the parents. The parents may opt to invite an "advocate," an individual who may be a seasoned parent or a professional who assists parents in ensuring

that their child receives all services to which he or she is entitled to, given the type and degree of delay or disability. The change in terminology from IFSP in the younger child to IEP after age 3 reflects a transition in focus from the family to the child. All children will receive structured educational activities in a classroom environment as guided by the child's IEP. Additionally, if appropriate and included in the IEP, the child will receive speech, occupational, and/or (but less frequently) physical therapy. More often than not, the therapy will be "consultative" rather than "hands-on" or "direct." This means that the therapists will periodically evaluate the child's progress, update goals and objectives, and provide the teachers and/or aides with individualized strategies to promote skill attainment.

**Elementary School.** As the child ages and becomes eligible for "elementary school," another evaluation will be conducted to determine continued eligibility for special education services. In addition to describing the type, intensity, and frequency of the services recommended, the team will also determine the best setting(s) for delivery of the services. The IDEA of 1990 and its subsequent re-authorizations in 1997 and 2004 state that the child should receive educational services in the "least restrictive environment." That is to say that the child should receive services in a regular classroom environment as much as possible without compromising delivery of the special services needed to ensure success. The extent of inclusion may depend on the level of MR, the severity of associated comorbid conditions, and especially, maladaptive behaviors (aggression, self-injurious, etc.). It also depends on the school district's resources. Inclusion in regular classes in children with milder MR can often be accomplished with a teacher aide who is shared among several students with special needs. During certain class periods, a special education teacher might provide individualized instruction in the regular classroom either directly to the student and/or consultation to the aide, who will then in turn provide the direct help. When service delivery is not feasible in the regular classroom due to severe degrees of cognitive impairment or behavior problems, the child might attend a "resource classroom" for one or more academic subjects.

**High School.** As the child with more mild forms of MR approaches adolescence, skills needed for successful independent living and meaningful employment should be targeted. Public law dictates that these issues are addressed in the development of the IEP

when the teen approaches 16 years of age; hence, the document and plan may be renamed as an ITP or Individualized Transition Plan. The ITP is different from the IEP in that:

- The student is a member of the ITP team and helps, to the best of his or her ability, to determine the educational goals, objectives, services, and settings in which services will be delivered.
- The emphasis changes from academic to vocational services and from remediating deficits to fostering abilities. A vocational assessment is conducted to evaluate the teen's interests and strengths and to determine the services needed to promote independence in the workplace and the community. Depending on the individual's cognitive level, health condition, work habits, and behavioral challenges, preparation for one of the following types of employment is targeted:

**Competitive employment.** The individual is hired, trained, and compensated in a manner similar to those not disabled. The job may include unskilled, semiskilled, or in some cases, even skilled duties. Work takes place in an integrated environment with minimal or intermittent supports.

**Supported employment.** The individual is hired to perform specific duties and is provided a job coach and/or environmental or schedule modifications that are necessary for success. As the individual masters the skills needed, coaching services are faded out. The individual earns competitive wages.

**Sheltered employment.** The individual works under constant supervision in a segregated setting. Often the work is contracted with local businesses. Examples range from silk screening T-shirts to assembling and sealing individual packets of plastic eating utensils, napkins, salt, and pepper for fast food carry-out restaurants. The individual may receive a weekly stipend (rarely consistent with minimum wage standards) or he may be compensated per piece.

Depending on the teen's employment goal, off-campus on-the-job training may be an option.

- The team will discuss goals for independent living after graduation and will recommend training needed to promote success. The teen's cultural,

Rights were not granted to include this figure in electronic media. Please refer to the printed journal.

**FIG 1.** Queen Shannon Jones proudly waves to the cheering crowd as she is crowned Homecoming Queen of Cy-Fair High School (3230 students). A representative from The Arc of the United States stated that event signifies that the current generation of high school students have a changed attitude about their peers with disabilities since they have been learning right alongside them. (KEVER, Jeannie, "Queen for Life." Houston Chronicle November 1, 2003; photo courtesy of Kevin Fujii.)

ethnic, linguistic, and economic characteristics should be always be considered.

- Representatives from adult-oriented disability agencies (state vocational rehabilitation agencies and/or Association of Retarded Citizens) are invited to attend the meeting and provide input and recommendations. Occasionally, these agencies may provide transition services before graduation.

In most states, children with MR may attend public school through at least 21 years of age. Longer years in formal school programs usually result in additional years in vocational training on campus or at off-campus sites.

Throughout the high school years, it is important that students be included in regular school social events and extracurricular activities. This is again dependent on the absence of any disruptive behaviors that may prevent such inclusion. Although students with MR often benefit from participation in specialized programs, such as Special Olympics, inclusion in regular athletic events, band, pep squad, and social events like proms and homecoming festivities alongside of normal peers is important. These opportunities improve self-esteem and self-worth as it did for one teen with Down syndrome (Fig 1), who was voted homecoming queen at a regular Houston high school. Finally, many states have developed self-advocacy and leadership

training opportunities for teens and young adults with disabilities.

### *Community Integration*

Although school activities occupy most of children's awake hours, recreational, scouting, and social activities outside of the school arena are also important in promoting community integration over the life span. As is the case in the educational arena, absence of maladaptive behaviors may be more important to the child's successful inclusion in these activities than level of MR. All persons, including individuals with disabilities, benefit from recreation and leisure activities. Opportunities include activities sponsored by community parks and recreation centers, city athletic leagues, church youth groups, art, drama, music, scouts, camping, travel, and visits to national and theme parks. As with typically developing children, those with MR will have particular interests and talents that are unique to the individual. Additionally, some syndromes are associated with unique abilities; for example, girls with Rett syndrome often demonstrate a strong affiliation for music, so much so that new information presented within a musical context is more easily learned than through traditional verbal means. (Note: Specific organizations discussed below are referenced in the Resource Guide in the Appendix.)

**Youth Groups.** Scout and other youth programs (eg, YMCA and YWCA) have accepted children with mild MR into their programs for many years. Inclusion of children with more severe forms of MR, especially those with maladaptive behaviors, has been more challenging and usually accomplished only with the assistance of volunteer "buddies" in innovative programs made possible by heroic efforts of parents and agency personnel. Activities sponsored by such programs during the school year can play a significant role in teaching appropriate social skills. Membership in one's church youth group is another venue for inclusion and opportunity for development of social, leadership skills, and lasting friendships. More importantly, it fosters spiritual development and provides assistance with prioritizing life goals. Churches (even within denominations) and other religious organizations differ in their degree of accommodations for children with special needs. As in all other areas of community integration, children with mild MR can often fully participate in most youth group activities

though their understanding of God and spirituality may be more concrete.

Electronic youth “gatherings” are becoming more and more popular among non-disabled teens. Best Buddies International attempts to link children and teens to other students with and without intellectual disabilities through the Internet to form online friendships through their Best Buddies’ e-Buddies program.<sup>92</sup> Finally, many states have developed self-advocacy and youth leadership training opportunities for teens with disabilities, including those with mild to moderate levels of MR. These often take place at college campuses with student mentors and also provide excellent opportunities for socialization.

**Camps.** Camping activities, especially sleep-away experiences, can be enormously valuable to all children. Several studies have found that weeklong residential camps contributed specific gains in self-reliance, communication, independence, and self-esteem that carried over into home and community settings.<sup>93,94</sup> The National Dissemination Center for Children with Disabilities (NICHD) publishes an annual guide with information about summer camps for children with all types of disabilities.<sup>95</sup> The American Camp Association accredits camps nationwide<sup>96</sup> and has promoted and required some degree of accessibility to children with disabilities. Children with mild MR experience few barriers to access to regular camps and need few, if any, accommodations.

**Physical Recreation.** According to the National Center on Physical Activity and Disability, regular physical activity may improve health, enhance social behavior, and build self-esteem in children with developmental disabilities.<sup>97</sup> Students with MR will often participate in specialized athletics such as Special Olympics, a well-established, international program that provides year-around training and sports competition in a variety of summer and winter sports. Although competitively focused, athletes of all ability levels are encouraged to participate. These programs not only promote physical health and fitness, they also improve self-esteem and self-worth and provide opportunities for socialization and leadership.<sup>98</sup> KEEN (Kids Enjoy Exercise Now Foundation) is a more recent opportunity for individuals with severe and profound intellectual and developmental disabilities ages 5 to 30 years to enjoy recreational and social activities such as tennis, swimming, basketball, soccer, and fitness training. However, unlike competitive

sports like Special Olympics, the emphasis is on having fun and building self-esteem.<sup>99</sup>

**Art, Music, Dance, and Theater.** Many children with MR may enjoy participating in the “fine arts” and more opportunities are becoming available to individuals with a wide range of disabilities. VSA arts was founded in the 1970s to ensure that people with disabilities have full inclusion in the arts. VSA arts is a worldwide nonprofit endeavor that serves over 5 million people annually through programs that include training institutes, artist-in-residence projects, arts camps, and an awards program to recognize emerging artists. Services vary among local VSA affiliates.<sup>100</sup>

**Hobbies.** Just like typically developing children, those with MR may enjoy gardening, crafts, cooking, or other activities around the house. Deficits in reading, dexterity, memory, and processing skills may be barriers for some of these activities. Innovative solutions have been found in many cases; for example, two cookbooks target individuals who are nonreaders and utilize pictures and simple text: *Cooking Made Easy* by Eileen Laird<sup>101</sup> and *Visual Recipes: A Cookbook For Non-Readers* by Tabitha Orth.<sup>102</sup> These activities, besides being fun, serve as another means of helping individuals with MR gain a sense of independence.

**Travel.** Travel is another form of recreation and leisure; unfortunately, family vacations can quickly turn into a nightmare if parents do not research available accommodations nor prepare adequately for the trip. Theme parks offer another recreational outlet. Sometimes children with MR, especially those with comorbid medical or physical disorders, receive special discounts and/or special attention in theme parks. Children with severe MR and behavior problems may be issued free “fast passes” to decrease waiting time and possible behavior outbursts. Parents are encouraged to inquire in advance about the possibility of facilitated access measures.

## *Adolescent and Young Adult Issues*

During adolescence, the pediatrician will need to address unique issues such as puberty, sexuality, and transition to adulthood. Once again maladaptive behaviors can be the most challenging barrier to the adolescent’s successful inclusion in social, work, and community settings. Teens with or without MR tend to experience emotional mood swings that sometimes result in inappropriate behavior. Individuals with MR, especially those with more severe MR, may be less

aware of others' opinions and less inhibited in public settings.

**Puberty and Sexuality.** Until recently, the issue of sexuality was rarely addressed in children with MR. The topic was considered "taboo" for fear that the mere mentioning of it would unleash inappropriate desires and behaviors. Recently numerous training programs have been developed to teach teens with MR appropriate behaviors using videos, comic book stories, and role playing and there is a growing body of literature addressing the topic<sup>103,104</sup> Sexuality encompasses more than genital sex and includes gender awareness, the need to be liked and accepted, displaying and receiving affection, feeling valued and attractive, and sharing thoughts and feelings. Sexuality is a basic human function and successful social relationships foster self-esteem and contribute to higher quality of life. Social development is chiefly experiential, and teens with MR, especially more severe forms of MR, may have fewer opportunities than other teens do.

Teens with severe MR, especially those with inappropriate behaviors, may experience difficulty in developing social relationships for many reasons: stigmatizing dysmorphic features, lack of awareness of social etiquette, inappropriate sexual behavior, comorbid medical or physical disabilities, and overprotection from parents. Thus, teens with MR may need formal training in mastering social greetings, telephone skills, and proper etiquette, including inhibition of sexual urges, in public settings.

Females with MR, like any female, should strive to become independent in self-care and hygiene; this may be more challenging in those with more severe levels of MR. Some females may never accomplish independence and may experience extreme anxiety and fear during menses when they are unable to comprehend the concept of periodic benign bleeding. Gynecological care likewise may be complicated by increased anxiety and lack of cooperation during a typical pelvic examination. Preparation aided by pictures or role playing, having a trusted caregiver present during the appointment, and the use of alternative positions other than pelvic stirrups may minimize fear and stress.

Parents of females with MR often express concerns about the possibility of pregnancy, the possibility of sexual abuse, and the efficacy of available birth control methods. They worry that their daughter would never be able to care for the child. For males, parents are concerned about protecting the male from the obligation of parental support that he would not likely

be able to fulfill. Finally, there is the fear that the parents themselves would be required to care for their grandchild by default. This issue involves many religious and ethical beliefs. Although sterilization was an option in the past, there are now many effective and reversible alternatives.<sup>105</sup> In addition to traditional 30-day-cycle oral contraceptives, long-term contraception is now available, which may be helpful in teens with limited cognition and motivation and/or with limited physical dexterity.<sup>106</sup> These include monthly intramuscular injections, implants that are effective up to 5 years, weekly applied transdermal patches, and long-term progestin-releasing intrauterine devices that can provide protection for several years. A decision to use these interventions is between the physician, the family, and the individual and must take into consideration possible side effects, especially when the individual is taking medications for treatment of comorbid disorders. Although legitimate parental concerns may be evident, the well-being and religious and ethical beliefs of the individual with MR should prevail in all decision-making.

**Transition to Adulthood.** Transition is defined as the movement from child-centered activities to adult-oriented activities. The major transition areas include (1) from school to the workplace; (2) from home to community living; and (3) from child- and family-centered pediatric care to adult-oriented health care. As noted above in the "Education" section, the school often plays an important role in preparing the child for work and community living through the development and implementation of the "Individualized Transition Plan."

The passage from childhood to young adulthood can be a time of internal turmoil. During this period, the adolescent is experiencing rapid changes in physical, cognitive, psychological, and social growth. Additionally, he or she must master decision-making, autonomy, and independence. Recently, teenagers have encountered greater challenges as they must navigate through additional obstacles representing the "new morbidities" (ie, substance abuse, gang violence, teen pregnancy, sexually transmitted disease, emotional disorders, and teen suicide). For some adolescents with MR, the journey is even more difficult as they face additional barriers imposed on them by cognitive, psychiatric, medical, and/or physical comorbid disorders.

Adolescents with mild MR are likely to proceed through this transition in a manner similar to those

without disabilities, albeit at a slower pace and with more deliberate training in certain areas. An older teen or young adult with mild MR can be expected to marry, parent children, and work in the community. However, comorbid disorders and behavior problems may limit success. Overprotective parents might also impede progress. Allowing teens to take steps toward living independently is crucial for success. Just as with typically developing teens, this journey is not without its setbacks and failures. Natural consequences of one's poor judgment must be experienced; however, the self-reflection and problem-solving that should follow in the wake of such an occurrence may need more facilitation from adults. Some degree of intervention and guidance during crises may be needed throughout the life span. Work opportunities may be limited to unskilled or semiskilled jobs that do not require a college degree. Technical school, on-the-job training, or the services of a temporary job coach may be needed for success.

Transition in teens with more severe forms MR may be much more challenging. Adults should understand that, to the degree they are capable, they have the right to choose the life they want and to seek the support needed to reach the outcomes they desire. Quality of life can be improved significantly by fostering independence at an early age and by teaching adolescents and their families the values of self-determination. However, teens with severe MR may not be able to fully transition to adult responsibilities. The AAMR<sup>6</sup> has outlined a system of support planning called an "Individualized Care Plan" that identifies ongoing needs and supports, including social supports, for such individuals in the home, workplace, and community. The system also calls for more intense temporary supports in times of stress or crisis. Recent laws promote living in the least restrictive and most inclusive community-based environments possible (Americans with Disabilities Act of 1990 [PL 101-336]; *Olmstead v. L.C.* Supreme Court decision [527 U.S. 581]). The main thrust of legislation has been to "normalize" the experiences of persons with MR as much as possible.

Once older teens with more severe forms of MR have graduated from high school, continued social and community involvement often becomes a formidable challenge. The regularly scheduled high school activities and dances tended to center around school events and athletics and are often no longer accessible to graduates. Without driver's licenses, it is more diffi-



**FIG 2.** Photo of king and queen. Queen Michelle Nixon and King Dustin Bianco enjoy an evening of triumph as they reign over the Mardi Gras Ball in Lake Charles, LA. (Color version of figure is available online.)

cult for them to be spontaneous or to attend community social events without supervision. Thus, they are at risk of becoming socially isolated. Fortunately, some sheltered employment agencies, in addition to offering work opportunities, also offer a broad array of activities for their clients to provide opportunities for socialization and expression of hobbies and talents. One center, Calcasieu Association for Retarded Citizens (CARC) in Louisiana, offers social activities several times a month that range from evening bowling leagues to formal balls and seasonal events such as the Mardi Gras Ball pictured in [Figure 2](#).

All teens (with or without cognitive deficits) automatically become their own legal guardians on their 18th birthday. If parents and the professionals working with the individual do not feel that she or he is capable of making responsible decisions, a formal team evaluation should be done to determine the need for guardianship. Procedures and terminology vary from state to state. It is wise to initiate the process in a nonrushed, proactive manner well before the teen's 18th birthday. If guardianship turns out to be in the individual's best interest, then legal services should be sought to help the parents navigate the legal/judicial system, to file a petition in a Probate Court, and to designate a "legal" guardian for the individual. Fees vary depending on the complexity of the decision-making and extent of legal services needed. The designee(s) might be one or both parents or an adult sibling, a relative, a family friend, or a professional. The guardian may be designated to make all decisions regarding the adult child's well-being; however, some

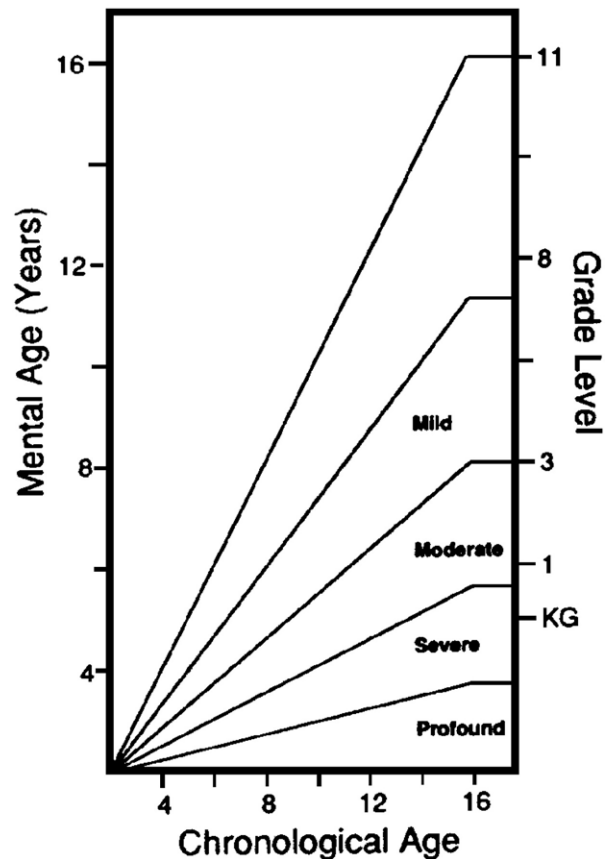
families may choose to divide decision-making responsibilities between the guardian and a conservator. A conservator chiefly manages the individual's financial affairs. When the adult has the intellectual capacity to make personal decisions but not complex financial ones (especially those relating to public supports), then only a conservator may be necessary.

Also at the 18th birthday, when their parents' incomes are no longer considered in the Supplemental Security Income (SSI) eligibility process, teens with severe disabilities, including severe MR, may qualify for SSI benefits. The teen must meet both disability and asset criteria. An employed adult with mild MR will not be eligible. Most individuals with severe MR will qualify for SSI; in most states, they will then also automatically become eligible for Medicaid benefits.

All older teens, including those with more severe forms of MR, should eventually transition from pediatricians to providers of adult health care.<sup>107</sup> There are fewer barriers for healthy adults with *mild MR*, especially when they receive health benefits from their employer and have no comorbid disorders. There are many barriers when individuals have *severe MR*, especially when complicated by medical disorders that heretofore were considered disorders of childhood. Although they will likely have adequate public insurance through Medicaid, access may still be a problem. Adult providers may be reticent to accept individuals with limited or no verbal skills, especially when there are complex comorbid problems. The internist or other provider of adult health care may also view the process as having a negative economic impact due to the additional care coordination efforts that may be needed and may be unwilling or unable to invest the time and financial resources into becoming knowledgeable and skillful in caring for this "new" adult disease.

### Prognosis

Prognosis relates to various aspects of life: longevity, health, adaptive functioning, ultimate academic achievement, employment, family life, community integration, and, ultimately, quality of life.<sup>90</sup> The degree of cognitive deficit contributes greatly to adult outcomes, especially in demarcating outcomes of individuals with mild versus severe MR. Due to variability among developmental and intelligence test scores in early childhood, predictions are difficult. In children with idiopathic mild MR, the predictive value of serial testing at young ages is enhanced when scores



**FIG 3.** Academic prognosis in mental retardation. Reprinted with permission from Accardo PM and Capute AJ. Mental retardation (Chapter 14). In: *Developmental Disabilities in Infancy and Childhood*. Baltimore: Paul Brookes Pub. Co.; 1996. p. 211.

are consistent throughout the elementary school years. Prognosticating is also easier when the child has a well-known genetic syndrome that is characterized by a consistent IQ range and for which long-term outcome studies exist.

Although intelligence is important in predicting ultimate academic skills (Fig 3), other factors more dramatically impact daily functioning and quality of life. These include coexisting behavioral, psychiatric, medical, and sensory disorders. Motivation and environmental factors such as societal barriers, past experiences in inclusive settings, and the individual's parents' parenting style (ie, overprotection versus promotion of independence) are important, especially in mild MR. Thus, very different outcomes can occur in individuals with similar IQs. With increasing age, social and adaptive skills become even more important in fostering successful functioning in the community.

As noted above, most individuals with MR have *mild* MR. In the absence of comorbid disorders, these individuals can be expected to have a normal life expectancy and a reasonably good quality of life.<sup>108</sup> They learn at one-half to two-thirds of normal velocity and usually achieve a 3rd to 6th grade reading level by late adolescence (Fig 3). During high school, they will often be enrolled in vocational classes; with good work habits they can be expected to become gainfully employed in competitive unskilled, semiskilled or, in some cases, skilled jobs. Unless there are comorbid disorders (health, physical, and/or psychiatric), they will often be self-supporting, marry, and parent children. As with parents with normal intelligence, they may benefit from periodic parent training and support, especially during stressful times.

Individuals with IQs in the middle range, such as those with Down syndrome, will learn at one-third to one-half velocity and achieve a 1st to 3rd grade reading level. They often live in group homes with supervision, although some adults live successfully on their own with the assistance of periodic “visitors” that help with shopping, balancing the checkbook, etc. Relatively few marry and parent children. Some work in supportive employment settings, but most work in supervised, sheltered workshops.

Environmental factors do not impact prognosis to the same extent in more *severe* forms of MR. The degree of disability is the chief limiting factor and thus outcomes are less optimistic. Those with IQ of 40 and below have a higher prevalence of medical comorbidity that often negatively impacts life expectancy. Those functioning in the higher range of this group may learn survival reading (ie, “stop” and “exit” signs). They usually attend “life skills” classes during their school years and will often require help with activities of daily living throughout their life. Severe cognitive and adaptive functioning deficits, especially when accompanied by challenging behaviors, often preclude work opportunities (even in sheltered workshops) and living in integrated community settings.

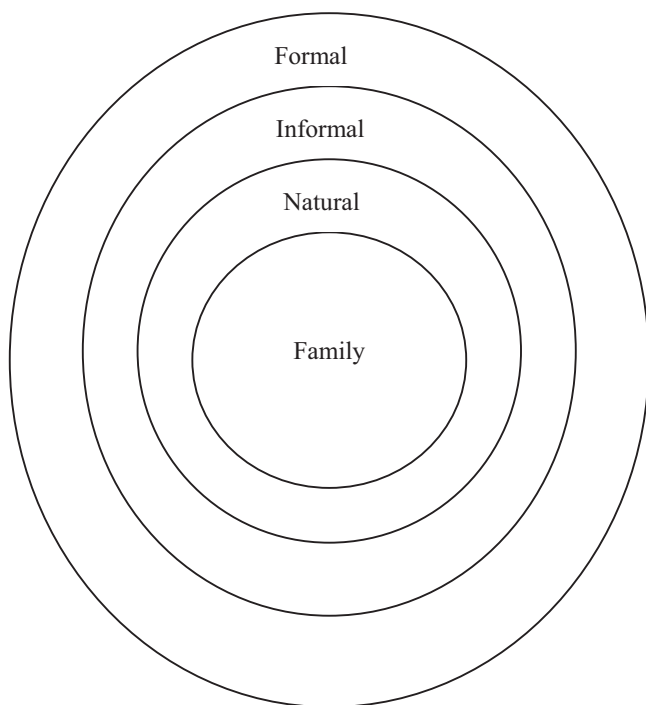
## Family Support

Parents of children with MR, like all parents, want to raise their child in the loving, nurturing environment of the home. Yet, unlike other families, parents of children with MR may be faced with prolonged care-giving and supervision responsibilities, especially

if the child has comorbid health care needs. Parents thus find their resiliency tested from the cumulative effect of minor day-to-day frustrations and during times of major life-status transitions or “critical periods.” Coping skills are often challenged, for example, when the child’s diagnosis is first made, when a younger sibling developmentally “passes up” the child with MR, or when the child with MR enrolls in “special” education. Later, critical periods may be triggered by the onset of puberty, graduation from high school, transition from pediatric to adult health care providers, or when teens are excluded from typical adolescent activities such as social/athletic events, dating, and driver’s education.

The birth of a child with a syndrome known to be associated with MR, or the discovery that a child has MR later in life, has a profound effect on siblings. Some evidence suggests that siblings of an affected child may be more sensitive, mature, and accepting of people with differences. On the other hand, younger siblings may harbor feelings of guilt because they were not born with a disability or resentment because of the parental attention that a brother or sister with a disability requires. Siblings are frequently called on to baby sit, which can become excessive. Parents may place an undue burden on older siblings to excel, and for some siblings, the pressure to please is often self-induced. If the child with MR is the older sibling, family dynamics are permanently altered when the younger sibling developmentally “passes up” his or her brother or sister with a disability. In other words, the nature of the sibling relationship may be positive or negative and, often, changes over time as the sibling learns to adapt and cope. The relationship may be influenced by a number of external factors, as well, including the type, quality, and availability of support services.<sup>109</sup>

Only 50 years ago, parents and siblings either had to raise the child with MR at home without any supports or consider a residential placement. Due in part to the efforts of disability advocates and some parents, attitudes slowly began to change and legislation was enacted that encouraged community supports and in-home care. Since the Americans with Disabilities Act in 1990, there has been a move toward de-institutionalization. In 2000, Healthy People 2010 established the goal of “zero children in congregate care facilities by 2010.”<sup>110</sup> The move has gained the backing of the AAP, which recently published the report, “Helping Families Raise Children with Special



**FIG 4.** Levels of support for families with children with mental retardation.

Health Care Needs at Home”.<sup>111</sup> Despite such legislative measures, it was not until 2001, for the first time in history, that funding for community support services exceeded funding for state institutions.<sup>112,113</sup>

To assist families raising their child with MR at home, ideally, families need access to three levels of support: natural, informal, and formal,<sup>114,115</sup> represented by three concentric circles as shown in Fig 4. At the innermost circle is the family’s *natural support system*, which includes those resources closest to the family such as spouses, immediate and extended family, friends, and the family’s faith community. These natural supports are often the family’s primary source for emotional and physical support. As families venture beyond their immediate circle of family or friends, they begin to utilize informal and formal supports. *Informal supports* include community-based services such as respite, child care, parent training, specialty clinics, social activities, and support groups that provide networking opportunities with other families of children with MR. Products and equipment that assist families with daily living activities and promote independence are, likewise, informal supports. Represented by the outermost circle, *formal supports* consist of state-administered and publicly funded programs

such as special education, accessible transportation, SSI benefits, Medicaid, and In-Home and Community-Based Waiver Services. This section on Family Support provides an overview of the three rings of supports available to families of children with MR.

### *Natural Supports*

Immediate family, friends, and neighbors are usually the primary assets on which parents depend, especially in the beginning. Families that are more mobile or that, for various reasons, have been “abandoned” by their extended families are at higher risk for inadequate natural supports. Religious organizations can be of tremendous support to these and all families. For example, some churches have developed specialized Sunday school programs for children with disabilities, including those with MR. A few provide Mother’s Day Out programs or other forms of relief for parents and welcome children with MR. Unfortunately, such children are often denied access to such programs, and parents may not be able to attend worship services, resulting in alienation from faith communities that may be otherwise a very important source of support.

### *Informal Supports*

As families move beyond their natural support system, either by choice or out of necessity when natural supports are not available, they begin to utilize informal supports. Community-based services and adaptive products help comprise an informal support system available to the family, siblings, and the child, respectively. An exhaustive discussion is beyond the scope of this article. A Resource Guide with a brief description of specific agencies, services, and products is appended to this article for the readers’ convenience. Specific organizations and products discussed in this section are referenced in the Guide.

***Services for the Family.*** While families of children with MR look to community-based services to supplement their natural supports, they may find that the depth and breadth of services vary, even within the same state or region. In rural locations, services are typically few and families may rely heavily on natural supports. Urban areas, on the other hand, tend to have a greater variety of programs.<sup>114</sup> At the same time, fees for services are not uniform. Some agencies offer services free of charge; others utilize a sliding fee based on the parents’ income to make it possible for all families to afford services, and other organizations charge only a minimal copay because of contractual

agreements with state agencies where services are subsidized with government grants.

**Parent Groups.** As parents search for information about their child's disability, they often turn to support groups formed at the local level to provide networking opportunities, offer guidance, and conduct advocacy at the state and national levels to improve public supports. Many of the formal disability organizations that exist today began as groups of parents that banded together informally to offer support and share information. Over time, these grassroots efforts evolved into national organizations. The Arc is one example. Founded in 1950 by a group of parents and others at a time when little was known about MR, today, the Arc has some 140,000 members with approximately 1000 chapters across the country.<sup>116</sup>

Today, national disability organizations engage in a broad range of disability-related activities. They disseminate information, generate public awareness, conduct regional and national conferences, provide opportunities for parent networking, publish newsletters and fact sheets, serve as a source for information referral, engage in advocacy efforts, and maintain a research registry. Many fund raise and those with ample resources often fund research. Some have state and local affiliates; however, programs and services may vary substantially among affiliates.

Some national organizations like the National Down Syndrome Society are disability-specific. Others, like the Sotos Syndrome Support Association, serve those with rare disorders. While organizations serving those with rare disorders may have fewer members and engage in fewer activities, parents routinely learn about respite services, adaptive sports, and equipment via these networks. Parents involved in these organizations often volunteer their time to increase public awareness about the disorder.

Finally, some organizations address issues that are important to families of children with all types of disabilities. Parent Advocacy Coalition for Educational Rights or PACER, for example, provides support and advocacy for families of children with all disabilities. The National Dissemination Center for Children with Disabilities (NICHCY) is a national information clearinghouse.<sup>117</sup> Located in every state, Parent Training and Information Centers provide training and disseminate information to parents and the professionals who work with children with MR. The aim of these centers is to ensure that parents are active collaborators, partners, decision-makers, and problem-

solvers alongside professionals and agency personnel in the care of their child with MR.<sup>114</sup>

**Conferences and Expos.** Disability organizations regularly host annual or biennial conferences at the regional, state, or national levels for parents, siblings, children with MR, and the professionals who serve them. In addition to providing opportunities to learn about the latest research results or therapeutic techniques, conferences commonly include parent networking opportunities. The World Congress on Disability and other national organizations conduct large disability expos. Workshops address a wide range of topics and vendors disseminate information about an array of products and services.

**Internet.** The internet has rapidly become an important tool to access disability information. Unfortunately, information found on many web sites is not peer reviewed; thus quality and validity may be questionable. Parents should be warned of this and advised to use caution in interpreting information from non-peer-reviewed sites, especially those that offer "cures." Rosenbaum and coworkers offer guidelines for parents using the internet as a source of information.<sup>118</sup> There are also reputable peer-reviewed sites that provide a scientific critique of alternative medicine interventions (eg, Consumer Federation of America: <http://www.quackwatch.com>). Small and large national disability organizations alike have created user-friendly web sites; many, but not all, are monitored by professional advisory councils. It is wise to advise parents to determine whether the site has indeed been reviewed by recognized experts in the field before relying on the information in the care of their child. Parents can also download e-newsletters and disability publications, obtain information about upcoming conferences, and link to related sites. Some web sites include real-time "ask the expert" sessions. Listservs, message boards, and electronic discussion groups enable parents to expand their peer support network across the globe. For those families without a computer or internet service at home, public libraries now commonly offer internet access.

**Respite.** Parents of all children benefit from an occasional break in the daily care-giving routine. Families of children with MR are faced with added care-giving and supervision responsibilities, especially if the child has complicated health care or behavioral needs. Parents often turn to extended family members to baby sit and provide a much needed break. If there are no relatives living nearby or if friends and family

are hesitant to provide care for a child with severe MR, parents (and sometimes entire families) may be at risk for burnout. To help these families recharge and face the care-giving role with renewed vigor, families need respite. Respite, the temporary care of a child with a disability by a trained provider for the purpose of providing relief to the family, has been documented as the service that families of children with MR report they need most.<sup>119-121</sup>

Parents seeking respite should be aware that there are a variety of respite models, and the availability and type of respite options vary within communities. Respite programs include center-based care, which utilizes trained caregivers and nurses to provide care in facility settings, or churches may partner with parent groups to offer faith-based respite. Some families may prefer to pay a trained provider to care for their child in the home. Although convenient, in-home respite is often costly, especially if nursing care is required. Family co-ops offer a more affordable alternative. In this case, families agree to “swap” care at no cost with other families. In extreme cases, parents may need an immediate break, or emergency respite, to cope and, occasionally, as a measure to prevent abuse.

**Child Care.** Parents seeking child care for a child with mild MR confront the usual daycare challenges that all parents face. Securing adequate, accessible, and affordable child care for a child with severe MR and comorbid behavior challenges and/or health care needs is a far greater challenge. The traditional neighborhood babysitter may lack the skills or comfort level to provide adequate care for the child. Despite ADA (1990) mandates, some child care centers remain ill-equipped or inappropriately staffed to accept children requiring specialized care. When child care is available, it is often expensive. The cost may far outweigh the potential earned income.<sup>122</sup> Even if both parents were employed in the workforce before the birth of the child, families routinely decide that one parent should quit work to stay home with the child with severe MR.

**Parent Support Groups.** Parent support programs provide parents with opportunities to meet other parents of children with MR to offer emotional support, share resource information, and exchange coping strategies. Some parent support groups recruit and match veteran parents of CWD with other parents in need of support. Often times, such groups will seek out and match parents of children with similar disabilities and family experiences. If the child has a rare disorder,

parents may feel particularly isolated from others. Some organizations utilize parent volunteers to reach out to other parents, offer emotional support and sometimes, to conduct gatherings. These programs are often particularly important to parents of a newly diagnosed child.

**Siblings.** Like parents, siblings of children with MR need interaction with other siblings. They also need information and, until recently, had fewer prospects for obtaining information.<sup>123</sup> The Sibling Support Project (SSP) of the Arc of the United States, created SibShops, which have been replicated in locations across the country to address sibling needs. SibShops convene school-aged siblings of children with disabilities, in a casual setting, for an afternoon of fun, activities, discussions, information exchange, and peer support under the direction of trained facilitators.<sup>124</sup> To enable siblings to connect with other brothers and sisters of children with disabilities (CWD) across the globe, SSP created SibKids and SibNet, listservs for young and adult siblings, respectively. Some disability-specific organizations, such as the Prader–Willi Syndrome Association (PWSA), are reaching out to siblings, too. PWSA established an eSupport group to enable siblings of children with PWS to connect with other siblings.<sup>125</sup>

**Adaptive Products.** Thus far, this overview of informal supports has focused on community-based services. Any discussion of informal supports must include information about adaptive products, however. Adaptive and specialty products play an important role in helping families with the care-giving responsibilities. In a study of parents in two states receiving cash subsidies, 62% reported that they purchased adaptive toys and 40% reported that they purchased diapers with the funds.<sup>126</sup>

**Toys.** All children benefit from toys and play. To ensure full access to play for the child with a disability, parents should select toys that complement the developmental level and abilities of the child.<sup>127</sup> Boehm<sup>128</sup> suggests that toys for the child with MR should motivate without being overly complex or frustrating. Caregivers may need assistance in identifying appropriate toys. The National Lekotek Center disseminates information about accessible play and has a toll-free Toy Resource Helpline to assist parents in the toy selection process.<sup>129</sup> *Exceptional Parent's* annual toy review is another source for information about toys for the child with MR.<sup>130</sup>

**Clothing.** Adaptive clothing products provide parents with relief from the daily care-giving activities and help promote the child's independence. Shirts and pants with Velcro® closures in place of buttons are a good example. The parent of a child with severe cognitive deficits may need to accompany the child to the restroom each time to assist with buttoning and zipping trousers. If the child wears adaptive pants with a Velcro® closure, he or she may be able to accomplish this daily-living skill without assistance. In some cases, the child with severe MR may never be toilet-trained and will require diapers into adulthood. Adult-size diapers can be expensive. Medicaid covers these types of expenses in some states. Special Clothes for Special Children and AbleApparel are just two of the many vendors that manufacture and sell adaptive clothing products and accessories.

**Adaptive Sporting Equipment.** Some children with MR have moderate balance impairments, but can enjoy riding a bicycle independently if it has a three-wheel design for added stability. Two- and three-wheel tandems provide support and the presence of an adult, if needed. Water-related products such as flotation vests offer access to pools and beaches. Of course, safety is always a priority when using adaptive sporting equipment as some individuals with MR may be less aware of danger.

### *Formal Supports*

Formal supports include publicly funded programs that provide financial assistance (ie, Supplemental Security Income benefits and Food Stamps, etc.), education, medical (Medicaid), vocational, and residential/living services. For the most part, publicly funded supports exist because of federal legislation, often promoted by the heroic efforts of parents and disability advocates. Implementation of these laws at the state level is largely influenced by budgetary restrictions. Some of these supports are available to ALL children, such as a free public education. Educational programs have already been discussed in the above section on Management and will not be addressed further. Other public benefits are need-based; access depends on financial need and/or severity of disability (mental retardation). Most individuals with mild MR will not meet the disability severity criterion for public benefits; additionally, if they are gainfully employed, they may not meet financial criteria.

**Financial Supports.** In 1974, SSI benefits (distributed by the Social Security Administration [SSA])

became available and represented the first support to families raising children with severe disabilities, including those with MR, at home. However, it was, and continues to be, a "needs-based" (ie, based on income and assets) benefit for *low-income* families.<sup>131</sup> Eligibility during childhood depends on a disability-severity criterion and a financial criterion for the family. If the child meets both criteria, the family will receive a monthly stipend; additionally, in most states, the child will automatically become eligible for Medicaid. The benefit amount has steadily increased over its 30-year existence. In 2005, the maximum monthly subsidy is \$564; however, the actual stipend may be less due to several factors. Qualification for any amount of stipend makes the child Medicaid-eligible. Older teens with severe MR may first become eligible for SSI (and Medicaid) benefits at the 18th birthday, when they are considered adults and their parents' income and financial assets are no longer considered in the eligibility process. Additionally, some families, depending on their financial circumstances, may also be eligible for Food Stamps and subsidized housing; these options are available to low-income families with typically developing children as well as to those with MR.

**Residential and Daily Living Supports.** In addition to supports provided through SSI, educational, and health care programs (Medicaid), families of children with MR, especially those with severe forms of MR, may be eligible for additional supports to assist them in raising their child. In the past, the only way a family could access these supports was to institutionalize their child. In the 1970s the vast majority of funding supported large state-operated institutions; however, legislation has subsequently shifted the preponderance of funding to community-based services<sup>113</sup> (J. Agosta, personal communication). Although funding for community-based supports has been increasing over the past three decades, states vary a great deal in the degree of funding allocated for these services. Some states have policies that guarantee that all children with MR will live in family settings, but many states still allocate large sums to institutional settings such as state facilities, nursing homes, and Intermediate Care Facilities for the Mental Retarded.

The most common funding mechanism for families for in-home supports is called "Home and Community-Based Waiver Services" or HCBS. HCBS funding to families are called "waiver options" because the family's income and assets are "waived," making access equitable across all income levels; eligibility

depends entirely on the severity of the child's disability and the circumstances it imposes on the family. However, many states have long waiting lists. States have developed different mechanisms to distribute their limited resources; some use a combination of options. The options include, but are not limited to, first come-first serve waiting lists; urgency of need; time-limited supports; and a lottery system. In any event, once a child becomes eligible for a funded slot, the family will be assigned a case-manager. Case-managers work collaboratively with the families and providers to design an annual service plan that includes choices from a menu of possible supports including, but not limited to, respite (in-home, center- or camp-based), medical equipment, home modifications for accessibility, and other needed supports. The child usually also becomes eligible for regular Medicaid, which in itself is a great support to most families.

In addition to HCBS waivers to support families raising their disabled child at home, many states provide direct cash subsidies that enable parents to purchase services from an approved menu of options similar to those listed above for the waiver system.<sup>122</sup> This funding strategy is sometimes also called a "voucher program." These services vary greatly from state to state and are very vulnerable to budget cuts. By 2002, 20 states had designed cash-subsidy or voucher programs that, of all mechanisms, provide the parents the highest degree of flexibility and control. Annual distributions in 2000 ranged from \$350 to \$8500 per family.<sup>122</sup> Often states have opted to provide families with mix of funding strategies to include both waiver options and cash subsidies. One funding stream may predominate over the other depending on state policy. For example, the state that provided the lowest annual cash subsidy to families also provided the highest level waiver funding (\$13,600 per family) in the same year. In the voucher program, parents themselves recruit, train, and enter into a formal contract with an individual (sometimes an extended family member) or a provider organization.

Because each state has organized its services and access mechanisms differently, pediatricians and families must learn their own state's idiosyncrasies to access supports by contacting the state or county offices of the Departments of Health and Human Services, Mental Health, and Mental Retardation or the state developmental disabilities organization. Additionally, local parent advocacy organizations, The

Arc, early intervention administrators, and/or school district special education coordinators are often knowledgeable about various programs and their respective eligibility requirements. The goal is to access sufficient services so that families are supported and do not feel the need for out-of-home placements. To further prevent this, some states have adopted a unique "support family system" similar to shared custody arrangements between divorced couples. The birth family recruits a support family from extended family members or from the community often with the assistance of a state-designated agency.<sup>132</sup> The two families plan in advance weekends and other times where each will care for the child, thus giving the birth family scheduled times of extended respite. In this way, costly institutionalization is aborted and the child enjoys the benefits of growing up in stable and nurturing family environments.

Unfortunately, out-of-home placements of children, especially fully grown teens with very severe MR and unmanageable behavior problems (ie, aggression or self-injurious behaviors) that threaten family functioning and/or the safety of siblings, still occur. In the past, large state-operated institutions were the only option. As the public became aware of the abuses and limitations of these facilities, the concept of community-based living options emerged. This reflects progress in that persons with severe MR can now live in architecturally "typical housing" in more community-based inclusive settings. Typically, three to six persons share a publicly subsidized home and the supervisory oversight of shift workers or full-time permanent "house parents." The degree of true community inclusion and "normalcy" among all the various group homes vary a great deal.

**Safeguarding Public Supports.** As noted above, eligibility for some public supports depend on the financial status of the family when the child with MR is less than 18 years old or on the income of the adult child after his or her 18th birthday. The child or adult may lose these supports if a well-meaning family member bequeaths a monetary gift. However, supports can be protected with a "Special Needs Will and Trust"; thus, this is an important consideration for families. A critical aspect of the will is a statement indicating that the inheritance is to be used only for items and services not covered by Medicaid, SSI, or other federal subsidies. Failure to include this statement will result in the loss of benefits until the amount equivalent to the value of the inheritance is "spent

down.” In some cases, the individual may be required to repay the government for services rendered in the past. Additionally, siblings’ shares of the inheritance may be at risk. The development of a Special Needs Will requires legal action. Various nonprofit and for-profit organizations are available to inform and guide parents.

## Conclusion

MR is one of the most common significant disabilities. Making a timely diagnosis of MR depends on a high index of suspicion, especially in a child who appears normal and demonstrates mild language delays. Diagnosis is a two-part process that includes the clinical diagnosis of MR based on DSM IV and/or AAMR criteria and a search for an etiology. Ideally it should also include a multidisciplinary approach in determining the level and kinds of supports that the individual will need over the life span. Management begins with prompt referral to an infant intervention program or, in older children, to a public education system. Children with MR should be cared for in the context of a medical home and receive ongoing quality health, dental, and mental health surveillance, especially when the MR is associated with comorbid conditions. It is important to consider the well-being of all family members and help them identify and access appropriate public and community supports when necessary. Regardless of the degree of MR, parents should be encouraged to promote independence to the maximum extent possible throughout all stages of development. They should also begin long-term financial planning early in the child’s life that will protect the child’s entitlement to public supports as adults. The pediatrician plays important roles in recognition and diagnosis, promoting health and independence, preventing secondary disabilities, supporting both parents and siblings, and, finally, transitioning the adolescent to adult systems of care.

## Appendix 1: Resources for Families of Children and Adults with Mental Retardation

This guide is a helpful, although not exhaustive, list of informal and formal supports. *Exceptional Parent’s* Annual Resource Guide offers regularly updated information.

## Organizations for Children and Adults with Mental Retardation

### ***American Association on Mental Retardation (AAMR)***

444 N. Capitol Street, N.W., Suite 846  
Washington, DC 20001-1512  
Phone: (202) 387-1968 Toll-Free Phone: (800) 424-3688  
Fax: (202) 387-2193  
Web Site: [www.aamr.org](http://www.aamr.org)

AAMR conducts advocacy, sets policy, and is a leader in defining mental retardation. AAMR publishes *Mental Retardation: Definition, Classification, and Systems of Support* and offers training in the definition and classification system.

### ***The Arc of the United States***

1010 Wayne Avenue, Suite 650, Silver Spring, MD 20910  
Voice(301): 565-3842  
Fax: (301) 565-5342  
E-mail: [info@thearc.org](mailto:info@thearc.org)  
Web Site: [www.thearc.org/](http://www.thearc.org/)

The Arc (formerly the Association for Retarded Citizens) promotes inclusion and conducts advocacy on behalf of persons with mental retardation and their families while fostering research and education to prevent mental retardation in young children.

### ***The Chromosome 18 Registry and Research Society***

7155 Oakridge Drive, San Antonio, TX 78229  
Phone/Fax: (210) 657-4968  
Web Site: [www.chromosome18.org](http://www.chromosome18.org)

Comprised primarily of parents of individuals with chromosome 18 abnormalities, the Society assists affected individuals with overcoming barriers, funds research, and conducts educational efforts to promote understanding of the various syndromes.

### ***Cornelia de Lange Syndrome Foundation, Inc. (CdLS-USA)***

302 West Main Street, Suite 100, Avon, CT 06001  
Phone: (860) 676-8166 or (860) 676-8255  
Toll-Free Support Lines (US only):  
(800) 223-8355 or (800) 753-2357  
Fax: (860) 676-8337  
E-mail: [info@cdlsusa.org](mailto:info@cdlsusa.org)  
Web site: [www.cdlsusa.org/](http://www.cdlsusa.org/)

CdLS-USA seeks to ensure accurate and early diagnosis of CdLS, provides family support services, disseminates information, generates public awareness, and supports research. Services include a toll-free help line, peer support, and annual conference.

### ***The President's Committee for People with Intellectual Disabilities (PCPID)***

The Aerospace Center  
370 L'Enfant Promenade S.W., Room 701  
Washington, DC 20447  
[www.acf.hhs.gov/programs/pcpid/index.html](http://www.acf.hhs.gov/programs/pcpid/index.html)  
Fax: (301) 317-5897 (To request PCID publications by fax)

The PCPID is a federal advisory committee that advises the President and Secretary of the Department of Health and Human Services on issues important to persons with mental retardation.

### ***Smith–Magenis Syndrome PRISMS, Inc. (Parents and Researchers Interested in SMS)***

P.O. Box 741914, Dallas, TX 75374-1914  
Phone: (972) 231-0035  
Fax: (413) 826-6539  
E-mail: [info@prisms.org](mailto:info@prisms.org)  
Web site: [www.prisms.org/home.htm](http://www.prisms.org/home.htm)

PRISMS provides information and support to families of individuals with Smith–Magenis Syndrome (SMS), while increasing awareness and understanding of SMS through partnerships with professionals. PRISMS coordinates a peer support program, maintains a parent listserv, hosts an international conference, and publishes a newsletter.

### ***Sotos Syndrome Support Association (SSSA)***

P.O. Box 4626, Wheaton, IL 60189  
Toll-Free Phone: (888) 246-7772  
International Phone: (630) 682-8815  
E-mail: [ssa@well.com](mailto:ssa@well.com)  
Web Site: [www.well.com/user/sssa](http://www.well.com/user/sssa)

SSSA seeks to increase understanding of Sotos Syndrome through public awareness and an annual conference while offering opportunities for peer and professional support.

### ***Organizations Offering General Advice for All Disabilities***

#### ***National Association of State Boards of Education***

277 South Washington Street, Suite 100  
Alexandria, VA 22314  
Phone: (703) 684-4000  
Fax: (703) 836-2313  
Web Site: [www.nasbe.org](http://www.nasbe.org)

NASBE works to strengthen state leadership in educational policymaking, promote excellence in the education of all students, advocate equality of access to educational opportunity, and assure continued citizen support for public education.

#### ***National Dissemination Center for Children with Disabilities (NICHCY)***

Orally Box 1492, Washington, DC 20013

Voice/TTY: (202) 884-8200  
Voice/TTY: (800) 695-0285  
Fax: (202) 884-8441  
E-mail: [nichcy@aed.org](mailto:nichcy@aed.org)  
Web Site: [www.nichy.org/](http://www.nichy.org/)

NICHCY is a national clearinghouse that specializes in disseminating information about disability and education issues for children and youth ages birth to 22.

#### ***National Organization for Rare Disorders (NORD)***

55 Kenosia Avenue, P.O. Box 1968  
Danbury, CT 06813-1968  
Voice: (203) 744-0100 (Voice) TDD: (203) 797-9590 (TDD)  
Toll-Free Voicemail Only: (800) 999-6673  
Fax: (203) 798-2291  
E-mail: [orphan@rarediseases.org](mailto:orphan@rarediseases.org)  
Web Site: [www.rarediseases.org/](http://www.rarediseases.org/)

NORD is composed of national health organizations, researchers, physicians, and individuals dedicated to the interests of people with rare debilitating disorders. NORD provides information and referral and offers a program that links individuals with the same or similar illnesses.

#### ***PACER Center***

Technical Assistance Alliance for Parent Centers (Formerly  
Technical Assistance for Parents Program)  
National Technical Assistance Center  
8161 Normandale Boulevard, Minneapolis, MN 55437-1044  
Voice: (952) 838-9000  
Toll-Free: (888) 248-0222  
TTY: (952) 838-0190  
Fax: (952) 838-0199  
E-mail: [alliance@taalliance.org](mailto:alliance@taalliance.org)  
Web Site: [www.taalliance.org/](http://www.taalliance.org/)

The Technical Assistance Alliance for Parent Centers project provides technical support for the development, coordination, and assistance of Parent Training and Information Centers and Community Parent Resource Centers. The Alliance national office disseminates information and conducts a national conference. Regional Center services include an annual regional conference, other training programs, publications, and site visits.

#### ***Beach Center on Disability***

University of Kansas Haworth Hall  
Lawrence, KS 66045-7534  
Phone (913) 864-7600  
E-mail: [beachcenter@ku.edu](mailto:beachcenter@ku.edu)  
Web Site: [www.beachcenter.org/](http://www.beachcenter.org/)

The Beach Center is an international research, teaching, learning, and service center funded by the National Institute on Disability and Rehabilitation Research.

***DisabilityInfo.gov*** [DisabilityInfo.gov](http://DisabilityInfo.gov) is a comprehensive, interagency web site that serves as a single point of access to the

federal government's information and resources for individuals with disabilities.

### ***Project DOCC (Delivery of Chronic Care)***

National Contact: Nancy Hoffman  
Phone: (212) 494-0746  
E-mail: [projectdocc@aol.com](mailto:projectdocc@aol.com)

Project DOCC is a training program founded by three mothers of CWD that emphasizes the need for a partnership between parents and health care professionals in caring for the child.

### ***Exceptional Parent Magazine (EP)***

65 East Route 4, River Edge, NJ 07661  
Toll-Free Phone: (800) 372-7368  
Fax: (201) 489-0074  
Web Site: [www.eparent.com/](http://www.eparent.com/)

*Exceptional Parent* is a magazine for parents and professionals involved in the care and education of children with disabilities. *EP* publishes an Annual Resource Guide and an Annual Toy Review.

### ***Maternal and Child Health (MCH) Library and National Technical Assistance Center for Children's Mental Health at Georgetown University***

[www.mchlibrary.info/knowledgepaths/kp\\_community.html](http://www.mchlibrary.info/knowledgepaths/kp_community.html)

The MCH Library partnered with the National Technical Assistance Center for Children's Mental Health at Georgetown University to create a knowledge path to help families and providers access resource information.

### ***Parent, Peer, and Sibling Support Groups***

#### ***National Down Syndrome Congress (NDSC)—Parents***

1370 Center Drive, Suite 102, Atlanta, GA 30338  
Phone: (770) 604-9500  
Toll-Free Phone: (800) 232-NDSC  
E-mail: [info@ndscenter.org](mailto:info@ndscenter.org)  
Web Site: [www.ndscenter.org/](http://www.ndscenter.org/)

NDSC promotes the interests of persons with Down syndrome through information dissemination, advocacy, parent support, and public awareness. NDSC also hosts an annual convention.

#### ***Parent to Parent—USA (P2P-USA)—Parents***

Web Site: [www.p2pusa.org/](http://www.p2pusa.org/)

P2P USA is an alliance of statewide Parent to Parent programs that matches trained, veteran parents of cww with other parents of cww for peer support. P2P hosts a biannual international conference.

#### ***Parents Helping Parents (PHP)—Parents***

3041 Olcott Street, Santa Clara, CA 95054

Phone: (408) 727-5775  
Fax: (408) 727-0182  
E-mail: [info@php.com](mailto:info@php.com)  
Web Site: [www.php.com](http://www.php.com)

A nonprofit resource center, PHP's services include peer support programs, education and advocacy training, disability awareness workshops, information dissemination, a resource library, support group sessions, and an assistive technology demonstration laboratory.

#### ***Voice of the Retarded (VOR)—Parents***

5005 Newport Drive, Suite 108, Rolling Meadows, IL 60008  
Phone: (847) 253-6020  
Fax: (847) 253-6054  
E-mail: [vor@compuserve.com](mailto:vor@compuserve.com)  
Web Site: [www.vor.net/](http://www.vor.net/)

A national nonprofit comprising families, professionals, and organizations, VOR seeks to ensure that children with MR have access to residential services including community- and facility-based care. VOR conducts advocacy and holds an annual meeting and Washington Initiative.

#### ***The National Father's Network—Fathers***

Washington State Fathers Network  
Kindering Center  
16120 N.E. Eighth Street, Bellevue, WA 98008  
Phone: (425) 747-4004 Extension 4286  
Fax: (425) 747-1069 (Fax)  
E-mail: [cmorris@fathersnetwork.org](mailto:cmorris@fathersnetwork.org)  
Web Site: [www.fathersnetwork.org/](http://www.fathersnetwork.org/)

The Father's Network is a national nonprofit that advocates for and provides support to fathers of CWD and professionals who serve them. FN develops curriculum to increase involvement of fathers, conducts conferences, and offers mentoring programs.

#### ***Youth Leadership Forum for Students with Disabilities (YLF)—Peers***

Office of Disability Employment Policy (ODEP)  
1331 F. Street, N.W., Washington, DC 20004  
Phone: (202) 693-7880  
Fax: (202) 693-7888  
E-mail: [epstein.alicia@dol.gov](mailto:epstein.alicia@dol.gov)  
Web Site: [www.dol.gov/odep/](http://www.dol.gov/odep/)

Developed by the California Governor's Committee for Employment for Disabled Persons and replicated in several states, the YLF is a free 4-day career and leadership training program for junior and high school students with disabilities.

#### ***Best Buddies—Peers***

100 SE Second Street, Suite 2200, Miami, FL 33131  
Phone: (305) 374-2233 (Phone)  
Toll-Free Phone: (800) 89-BUDDY

Fax: (305) 374-5305  
Web Site: [www.bestbuddies.org/](http://www.bestbuddies.org/)

Best Buddies is an international nonprofit committed to providing supportive employment opportunities and promoting friendships between persons with intellectual disabilities and their peers.

### ***Project Cope—Peers***

Westchester Institute for Human Development  
Cedarwood Hall Room 338, Valhalla, NY 10595-1689  
Phone: (914) 493-5202  
E-mail: [projectcope@nymc.edu](mailto:projectcope@nymc.edu)  
Web Site: [www.nymc.edu/wich/projectcope/pc/](http://www.nymc.edu/wich/projectcope/pc/)

Project Cope is a national clearinghouse that provides resources to assist children with special needs and other vulnerable populations in the wake of a disaster.

### ***Sibling Support Project (SSP) of The Arc of the United States—Siblings***

6512 23rd Ave N.W., #213, Seattle, WA 98117  
Phone: (206) 297-6368  
Fax: (509) 752-6789  
E-mail: [donmeyer@siblingsupport.org](mailto:donmeyer@siblingsupport.org)  
Web Site: [www.thearc.org/siblingsupport/](http://www.thearc.org/siblingsupport/)

Founded by Donald Meyer, the SSP is a national effort that provides support and information to siblings of persons with disabilities while helping caregivers to understand the unique issues facing siblings.

### ***Respite***

#### ***ARCH National Respite Network and Resource Center***

A Service of the Chapel Hill Training-Outreach Project, Inc.  
800 Eastowne Drive, Suite 105, Chapel Hill, NC 27514  
Phone: (919) 490-5577  
Fax: (919) 490-4905  
Web Site: [www.archrespite.org/](http://www.archrespite.org/)

ARCH is a national center that offers training and technical assistance to families, providers, and state entities while conducting program evaluation and research in its efforts to create a nationwide system of quality respite services. Through its National Respite Locator Service, the ARCH helps families and professionals locate respite programs.

### ***Adaptive Clothing, Toys, and Equipment***

#### ***AbleApparel***

2121 Hillside Avenue, New Hyde Park, NY 11040  
Phone: (516) 873- 6552  
Fax: (516) 248-7308  
E-mail: [sales@ableapparel.com](mailto:sales@ableapparel.com)  
Web Site: [www.ableapparel.com/](http://www.ableapparel.com/)

AbleApparel sells adaptive products including pants, outerwear, bibs, and accessories.

### ***Special Clothes***

P.O. Box 333, Harwich, MA 02645  
Phone, TDD, and Fax: (508) 385-9171  
E-mail: [specialclo@aol.com](mailto:specialclo@aol.com)  
Web Site: [www.Special-Clothes.com/](http://www.Special-Clothes.com/)

Special Clothes for Special Children retails adaptive clothing for toddlers through young adults with disabilities.

### ***Tranquility Incontinence Products***

Principle Business Enterprises  
Pine Lake Industrial Park, Dunbridge, OH 43414  
Phone: (800) 467-3224  
Fax: (419) 352-8340  
Web Site: [www.tranquilityproducts.com/](http://www.tranquilityproducts.com/)

Tranquility products include briefs, pads, and liners. Free trial-sized sample product offer is available in the continental US.

### ***National Lekotek Center—Toys***

3204 W. Armitage Avenue, Chicago, IL 60647  
Phone: (773) 276-5164 TTY: (773) 276-8633  
Fax: (773) 276-8644  
Lekotek Toy Resource Helpline (800) 366-PLAY  
E-mail: [lekotek@lekotek.org](mailto:lekotek@lekotek.org)  
Web Site: [www.lekotek.org/](http://www.lekotek.org/)

Lekotek and its nationwide network of play centers disseminate information about accessible play and offer assistance in toy selection through a toll-free toy help-line. Other services include toy lending libraries, monthly play sessions, and interactive computer play programs.

### ***Sports, Camps, Recreation, and Travel***

#### ***Special Olympics***

1133 19th Street, N.W., Washington, DC 20036  
Phone: (202) 628-3630  
Fax: (202) 824-0200  
E-mail: [infor@specialolympics.org](mailto:infor@specialolympics.org)  
Web Site: [www.specialolympics.org/](http://www.specialolympics.org/)

An international organization, Special Olympics provides children and adults with intellectual disabilities the opportunity to participate in year-around sports training and competition.

#### ***Kids Enjoy Exercise Now (KEEN) Foundation, Inc.***

P.O. Box 341590, Bethesda, MD 20827-1590  
Phone: (301) 770-3200  
E-mail: [info@keenusa.org](mailto:info@keenusa.org)  
Web Site: [www.keenusa.org/](http://www.keenusa.org/)

Located in Washington, DC, Kansas City, and Chicago, KEEN enables individuals with severe mental and physical disabilities to participate in inclusive sports and recreational and social activities.

### ***Miracle League Association***

1506 Klondike Road, Suite 105, Conyers, GA 30094  
Phone: (770) 760-1933  
Fax: (770) 483-1223  
Web Site: [www.miracleleague.com/](http://www.miracleleague.com/)

Miracle League is an adaptive baseball league for children with disabilities. Volunteer “buddies” assist the players.

### ***American Camping Association (ACA)***

5000 State Road 67 North, Martinsville, IN 46151-7902  
Phone: (765) 342-8456  
Web Site: [www.acacamps.org/](http://www.acacamps.org/)

The ACA is a national organization of camp professionals dedicated to ensuring quality camping programs. ACA maintains a database of accredited camps serving children with disabilities

### ***Access to Recreation***

8 Sandra Court, Newbury Park, CA 91320  
Toll-Free Phone: (800) 634-4351  
E-mail: [dkrebs@accessr.com](mailto:dkrebs@accessr.com)  
Web Site: [www.accessr.com/](http://www.accessr.com/)

Access to Recreation retails adaptive equipment for daily living as well as for sports and recreational activities.

### ***Discover Camp***

Web Site: [www.ncaonline.org/discover/](http://www.ncaonline.org/discover/)

*Discover Camp* is a web site and booklet with practical information for parents choosing a camp program for their child with a disability for the first time.

### ***National Park Service***

U.S. Department of the Interior  
Toll-Free Phone: (888) GO-PARKS  
E-mail: [parkspass@nationalparks.org](mailto:parkspass@nationalparks.org)  
Web Site: [www.buy.nationalparks.org/golden.asp](http://www.buy.nationalparks.org/golden.asp)

The Golden Access Passport is available to US citizens with a disability. The pass provides free lifetime entrance into national parks and other select locations.

### ***Arts and Hobbies***

#### ***Partners for Youth with Disabilities (PYD)***

Access to Theater (ATT)  
95 Berkeley Street, Suite 109, Boston, MA 02116  
Phone: (617) 556-4075 TTY: (617) 314-2989  
Fax: (617) 556-4074  
E-mail: [mgallagher@pyd.org](mailto:mgallagher@pyd.org)  
Web Site: [www.pyd.org/](http://www.pyd.org/)

The Access to Theater (ATT) program engages youth with and without disabilities from the Boston area ages 13 to 24 to work collaboratively to create theatrical productions.

### ***VSA arts (formerly Very Special Arts)***

1300 Connecticut Avenue, Suite 700, Washington, DC 20036  
Voice: (202) 628-2800 TDD: (202) 737-0645  
Toll Free: (800) 933-8721  
Fax: (202) 737-0725  
E-mail: [info@vsarts.org](mailto:info@vsarts.org)  
Web Site: [www.vsarts.org/](http://www.vsarts.org/)

VSA arts is an international nonprofit dedicated to ensuring that persons with disabilities have the opportunity to learn about, take part in, and enjoy the arts.

### ***Cooking Made Easy***

Eileen Laird  
Orally Box 2117, Boone, NC 28607-2117  
E-mail: [eileen@cookingmadeeasy.org](mailto:eileen@cookingmadeeasy.org)  
Web Site: [www.apptechnc.net/~cme/homegt.htm](http://www.apptechnc.net/~cme/homegt.htm)

*Cooking Made Easy* is an adaptive recipe book created especially for individuals with disabilities.

### ***Visual Recipes: A Cookbook For Non-Readers***

DRL Books  
Toll-Free Phone: (800) 853-1057  
Online Ordering: [www.drlbooks.com/](http://www.drlbooks.com/)

*Visual Recipe* is a cookbook for nonreaders created by the mother of a child with autism.

### ***Advocacy and Legal Assistance***

#### ***Family Voices, Inc.***

2340 Alamo SE, Suite 102, Albuquerque, NM 87106  
Phone: (505) 872-4774  
Toll Free: (888) 835-5669  
Fax: (505) 872-4780  
E-mail: [kidshealth@familyvoices.org](mailto:kidshealth@familyvoices.org)  
Web Site: [www.familyvoices.org/](http://www.familyvoices.org/)

A national, grassroots effort comprising families of cwd, friends, professionals, and interested persons, Family Voices ensures that its members have a voice in health care policy discussions and decision-making about the health care of children with disabilities.

#### ***The George Washington University HEATH Resource Center***

2121 K Street, N.W., Suite 220, Washington, DC 20037  
Voice/TTY: (202) 973-0904  
Toll-Free Phone: (800) 544-3284  
Fax: (202) 973-0908  
E-mail: [askheath@gwu.edu](mailto:askheath@gwu.edu)  
Web Site: [www.heath.gwu.edu/](http://www.heath.gwu.edu/)

A national clearinghouse, the HEATH Resource Center collects and disseminates information on postsecondary training and education for persons with disabilities.

## ***Disability Rights Education and Defense Fund (DREDF)***

2212 Sixth Street, Berkeley, CA 94710  
Voice/TTY: (510) 644-2555  
Fax: (510) 841-8645  
E-mail: [dredf@dredf.org](mailto:dredf@dredf.org)  
Web Site: [www.dredf.org/](http://www.dredf.org/)

A national disability rights law and policy center founded by parents of cwd and self-advocates, DREDF offers training, legal support and advocacy, and analysis of policy questions.

## ***National Association of Protection and Advocacy Systems, Inc. (NAPAS)***

900 Second Street, N.E., Suite 211, Washington, DC 20002  
Phone: (202) 408-9514 TTY (202) 408-9521  
Fax: (202) 408-9520  
E-mail: [info@napas.org](mailto:info@napas.org)  
Web Site: [www.napas.org/](http://www.napas.org/)

NAPAS provides training, technical support, and leadership to protection and advocacy systems and client assistance programs (congressionally mandated disability rights agencies that provide advocacy services and legal representation for persons with disabilities).

## ***US Department of Justice***

Civil Rights Division  
950 Pennsylvania Avenue, N.W.  
Disability Rights Section—NYAV  
Washington, DC 20530  
Voice: (800) 514-0301 TTY: (800) 514-0383  
Web Site: [www.ada.gov/](http://www.ada.gov/)

The Department of Justice provides technical assistance and disseminates information about the ADA, standards for accessible design, and related topics through a 24-hour information line, web site, and publications.

## ***Guardianship, Special Needs Wills, Estate Planning, and Life Insurance***

### ***Disabled and Alone***

Life Services for the Handicapped, Inc.  
61 Broadway, Suite 510, New York, NY 10006  
Phone: (212) 532-6740  
Toll-Free Phone: (800) 995-0066  
Fax: (212) 532-3588  
E-mail: [info@disabledandalone.org](mailto:info@disabledandalone.org)  
Web Site: [www.disabledandalone.org/](http://www.disabledandalone.org/)

Life Services is a nonprofit that assists families with long-term care planning for family members with a severe disability.

## ***MassMutual SpecialCare<sup>sm</sup> Program—Estate Planning, Life Insurance***

SpecialCare<sup>sm</sup> Program, C301  
MassMutual Financial Group  
1295 State Street, Springfield, MA 01111-0001  
Toll-Free Phone: (800) 272-2216  
Web: [www.massmutual.com/mmfg/prepare/specialcare/index.html](http://www.massmutual.com/mmfg/prepare/specialcare/index.html)

Through MassMutual's program, a financial planning professional certified in SpecialCare<sup>sm</sup> assists the family in planning for the future of the child with a disability.

## ***MetDESK®—Estate Planning, Life Insurance***

MetLife's Division for Special Kids  
(877) MET-DESK or 638-3375  
Web Site: [www.metlife.com/desk](http://www.metlife.com/desk)

MetDESK® utilizes specially trained financial service professionals to assist families in navigating the maze of legal and financial planning for the future of a child with special needs.

## ***Special Needs Advocate for Parents (SNAP)***

11835 W Olympic Boulevard, Suite 465, Los Angeles, CA 90045  
Phone: (310) 479-3755  
Toll-Free Phone: (888) 310-9889  
Fax: (310) 479-3089  
E-mail: [info@snapinfo.org](mailto:info@snapinfo.org)  
Web Site: [www.snapinfo.org/homehtml](http://www.snapinfo.org/homehtml)

SNAP is a national nonprofit that provides parents with information, education, advocacy, networking, and referral services in areas such as special needs planning and navigating the private medical insurance system.

## ***Special Needs Alliance (SNA)—Guardianship, Special Needs Wills, and Trusts***

Toll-Free Phone: 1 (877) 572-8472  
Web Site: [www.specialneedsalliance.com/](http://www.specialneedsalliance.com/)

SNA is a national network of lawyers specializing in Disability and Public Benefits Law. Families of cwd can locate a SNA lawyer using the toll-free number or by requesting information online.

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