
**Review**

**MENTAL RETARDATION: DEFINITIONS, ETIOLOGY, EPIDEMIOLOGY AND DIAGNOSIS**

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RESUMEN

El objetivo de este trabajo ha sido proporcionar una visión general de retraso mental, y de la discapacidad en el desarrollo que presenta una larga historia y, en ocasiones, polémica. En particular, se van a presentar las actuales definiciones, las causas de retraso mental, la información epidemiológica y los criterios de diagnóstico.

Palabras clave: discapacidad intelectual, estudio de revisión, definición, causas, epidemiología, diagnóstico.

ABSTRACT

The objective of this paper was to provide an overview of mental retardation, a developmental disability with a long and sometimes controversial history. In particular, the current definitions, causes of mental retardation, diagnostic criteria and epidemiological information will be presented.

Key words: intellectual disability, review study, definition, causes, epidemiology, diagnosis.
INTRODUCTION

Mental retardation (MR) is a genetic disorder manifested in significantly below average overall intellectual functioning and deficits in adaptive behaviour. Mental retardation is a particular state of functioning that begins in childhood and is characterized by decreased intelligence and adaptive skills and also is the most common developmental disorder (Bregman, 1991). MR retardation in young children is often missed by clinicians. The condition is present in 2 to 3 percent of the population, either as an isolated finding or as part of a syndrome or broader disorder (Daily, Ardinger & Holmes, 2000).

Causes of mental retardation are numerous and include genetic and environmental factors. In at least 30 to 50 percent of cases, physicians are unable to determine etiology despite thorough evaluation (Baird & Sadovnick, 1985). Diagnosis is highly dependent on a comprehensive personal and family medical history, a complete physical examination and a careful developmental assessment of the child. These will guide appropriate evaluations and referrals to provide genetic counselling, resources for the family and early intervention programs for the child (Rutter, 2006).

DEFINITIONS

An accurate and consistent definition of mental retardation is critical because of its impact on the prevalence, or count, of those with MR. However, despite the importance of consistency, MR is not always defined in the same way across research studies or service agencies, even within the same state (Koller et al., 1984; Borthwick-Duffy, 1994). While some definitions rely on IQ scores alone to classify individuals with MR, some only use adaptive behaviours for classification, and others include both IQ scores and measures of adaptive skills (Whitman et al., 1990; Borthwick-Duffy, 1994). In addition, many studies are based on broad categories of either severity (using labels such as mild, moderate, severe and profound MR) or etiology (utilizing the terms cultural/familial and organic MR).

The American Association on Intellectual and Developmental Disabilities (AAIDD, 2002) has defined MR as significant limitations both in intellectual functioning and in adaptive behavior, which covers many everyday social and practical skills. This disability originates before the age of 18. Intellectual limitations refer to an Intelligence Quotient (IQ) which falls two standard deviations below the population mean of 100 (<70), and adaptive functioning limitations refer to impairments in at least two out of ten skill areas (AAMR, 2000). MR is also defined in the Diagnostic and Statistical Manual of Mental Disorders, 4th edition (DSM-IV) by the American Psychiatric Association (APA). Similar to the AAMR definition, the DSM-IV has three diagnostic criteria for MR, including sub-average intellectual functioning (IQ < 70), impairments in adaptive functioning and onset before age 18 (APA, 1994).

Zigler and colleagues (1986; 1987; 1991) argue that an appropriate classification of MR employs both IQ score and etiology of the retardation. Consequently, they suggest categorizing MR into cultural/familial and organic groups, based on the presence or absence of a known organic etiology. This two-group approach is one of the most well documented distinctions in the mental retardation literature over the last century.

CAUSES OF MENTAL RETARDATION - ETIOLOGY

Approximately 3% of the population has an intelligence quotient (IQ) of less than 70, among whom a cause for the mental retardation can be established in less than half of all cases (Flint et al., 1995). The
prevalence of severe mental retardation is about 3 per 1.000 population and 30 per 1.000 for mild mental retardation (Harper, 1993).

A number of environmental, genetic or multiple factors can cause mental retardation. It is also believed that behavioural or societal factors such as poverty, malnutrition, maternal drug and alcohol use, as well as severe stimulus deprivation can contribute to MR (McLaren & Bryson, 1987). Unfortunately, in approximately 30 to 50 percent of cases, the etiology is not identified even after thorough diagnostic evaluation (Cury et al., 1997; Schaefer & Bodensteiner, 1992). Some persons have a congenital malformation of the brain; others had damage to the brain at a critical period in pre- or postnatal development. Acquired causes of retardation include near-drowning, traumatic brain injury and central nervous system malignancy. The most common cause of MR in industrialized nations is fetal alcohol syndrome with an incidence rate of 1 in 100 births. The second leading known cause of MR is Down syndrome, or trisomy 21, with an incidence rate of 1 in 800-1,000 births (Campbell, Morgan & Jackson, 2004).

3.1 Genetics conditions

A number of single-gene disorders result in mental retardation. Many of these are associated with atypical or dysmorphic physical characteristics (Sultana et al., 1995). Such conditions include fragile X syndrome, neurofibromatosis, tuberous sclerosis, Noonan's syndrome and Cornelia de Lange's syndrome (Baraitser & Winter, 1996; Jones & Smith, 1997).

As many as one fourth of persons with mental retardation have a detectable chromosome abnormality. Children with Down syndrome (trisomy 21) usually have highly recognizable physical characteristics, but features associated with other chromosomal abnormalities, such as Klinefelter's syndrome (47,XXY), may not be as obvious to family members or the physician (Batshaw & Perret, 1992) Other children may have a small deletion or duplication of a particular chromosome that is rarely reported; thus, the phenotype is still undetermined. Some chromosomal abnormalities are inherited from a parent but most occur de novo (McKusick, 1994). Many previously described clinical syndromes have been found to have an associated chromosomal abnormality (e.g., DiGeorge, Prader-Willi, Angelman and Williams syndromes). Other genetic conditions include Phelan-McDermid syndrome (22q13del), Mowat-Wilson syndrome, genetic ciliopathy (Badano et al., 2006). In the rarest of cases, abnormalities with the X or Y chromosome may also cause disability. 48, XXXX and 49, XXXXY syndrome affect a small number of girls worldwide, while boys may be affected by 47, XXY, 49, XXXXY, or 49, XYYYY (Moser, 1995).

3.2 Prenatal problems

Mental disability can result when the fetus does not develop inside the mother properly. Moreover, prenatal causes include congenital infections such as cytomegalovirus, toxoplasmosis, herpes, syphilis, rubella and human immunodeficiency virus; prolonged maternal fever in the first trimester; exposure to anticonvulsants or alcohol; and untreated maternal phenylketonuria (PKU) (Strømme & Hagberg, 2007). Complications of prematurity, especially in extremely low-birth-weight infants, or postnatal exposure to lead can also cause mental retardation (Piecuuch et al., 1997).

3.3 Perinatal problems

Perinatal causes involve late pregnancy (complications of pregnancy, diseases in mother such us heart and kidney disease and diabetes and placental dysfunction), during delivery (labour) (severe prematurity, very low birth weight, birth
3.4 Postnatal problems (in infancy and childhood)

Postnatal problems include infancy and childhood. It is involved brain infections such as tuberculosis, Japanese encephalitis, and bacterial meningitis. As well as head injury, chronic lead exposure, severe and prolonged malnutrition and gross understimulation (Leonard & Wen, 2002; Zoghbi, 2003).

3.5 Metabolic disorders

Metabolic disorders are another possible cause of mental retardation. In some cases (e.g., PKU, hypothyroidism), retardation is preventable with early treatment (Scrivener, 1995). Other disorders (e.g., mucopolysaccharidosis, sphingolipidoses) are less responsive to early intervention. Molecular medicine has made it possible to diagnose a number of conditions referred to as mitochondrial cell diseases (Dimauro & Moraes, 1993).

3.6 Exposure to certain types of disease or toxins

Diseases like whooping cough, measles, or meningitis can cause mental disability if medical care is delayed or inadequate. Exposure to poisons like lead or mercury may also affect mental ability (Aicardi, 1998; Daily, Ardinger & Holmes, 2000).

3.7 Iodine deficiency (cretinism)

Iodine deficiency affecting approximately 2 billion people worldwide is the leading preventable cause of mental disability in areas of the developing world where iodine deficiency is endemic. Iodine deficiency also causes goiter, an enlargement of the thyroid gland (Delange, 1994). Lack of adequate availability of iodine from the mother restricts the growth of the brain of the foetus, and leads to a condition called hypothyroidism. More common than full-fledged cretinism, as retardation caused by severe iodine deficiency is called, is mild impairment of intelligence (Gaitan & Dunn, 1992). Certain areas of the world due to natural deficiency and governmental inaction are severely affected. India is the most outstanding, with 500 million suffering from deficiency, 54 million from goiter, and 2 million from cretinism. Among other nations affected by iodine deficiency, China and Kazakhstan have begun taking action, whereas Russia has not (McNeil, 2006).

3.8 Malnutrition

Malnutrition is a common cause of reduced intelligence in parts of the world affected by famine, such as Ethiopia (Durkin et al., 2000; Wines, 2006).

DIAGNOSIS

The first and most important step in the diagnosis of mental retardation is to obtain a comprehensive patient and family history. Previous gynecologic and obstetric history may reveal infertility or fetal loss (Matson & Sevin, 1994). Assessment of maternal health status during pregnancy with the involved child should include questions regarding use of tobacco, alcohol and drugs (prescribed and illicit); lifestyle or other risks for sexually transmitted diseases; weight gain or loss; signs of infection; serious illness or injury; and surgery or hospitalization (Reiss, 1994; Szymanski, 1994).

To establish a knowledgeable baseline history of the child, the physician should obtain information regarding length of pregnancy, premature onset of labor or rupture of the membranes, duration and
course of labor, type of delivery and any complications (Kolevzon, Gross & Reichenberg, 2007). Apgar scores at one and (especially) five minutes should be reviewed, and birth weight, length and head circumference measurements obtained and plotted on appropriate growth charts. The parents should be asked about any illnesses, feeding or sleeping difficulties in the newborn period and problems with sucking or swallowing, as well as the baby's general disposition (Leonard & Wen, 2002). Extremes in infant temperament are often the first clue to an atypical course in child development (Kolevzon et al., 2007).

The systems review of the child should be complete, with special attention to growth problems, history of seizures, lethargy and episodic vomiting. A developmental screen should be used at all well-child visits to obtain information about the timing of the child's developmental milestones, any concerns by parents or caregivers and comparison of the child's developmental rate and pattern with those of siblings (Palmer & Capute, 1994). Specific questions about the child's current developmental abilities should be asked at each visit.

The Revised Denver Prescreening Developmental Questionnaire (Frankenburg et al., 1981) is a useful screening tool that parents can readily complete to help determine the need for further evaluation with the time-honored Denver Developmental Screening Test (Frankenburg & Dodds, 1967; Frankenburg & Dodds, 1990). Another practical and reliable tool with which to monitor development in infants is the Kansas Infant Development Screen (Holmes & Hassanein, 1982). The findings can be recorded and plotted just as with somatic growth charts and shared with parents. Other developmental screening tests are also available.

Delays in speech development are common and may become more obvious when contrasted with the speech development of a sibling. Inquiry should be made regarding concerns about hearing and vision (Van Naarden, Decoufle & Caldwell, 1999). One cannot overemphasize the importance of addressing concerns voiced by a parent about a child's development, behaviour and learning; because these expressed concerns accurately target the majority of children with developmental problems.

Information should be obtained about the family unit, parents' occupations and educational achievements, educational and developmental status of siblings, role of the patient in the family, discipline of the children and identity of the child's caregiver when the parents are not home (Daily, Ardinger & Holmes, 2000). Family history of fetal loss, mental retardation, severe learning problems, congenital abnormalities and unexplained childhood deaths, as well as other serious illnesses in first- and second-degree family members, should be elicited. A complete physical examination can begin with a review of growth curves since birth, if these are available. The head circumference should continue to be plotted. The examination should be thorough, with special attention to physical findings that are compatible with any risk factors obtained from the history.

The child should be examined closely for dysmorphic features or minor abnormalities, such as unusual eyebrow pattern, eyes that are widely or closely spaced, low-set ears or abnormal palmar crease patterns. Minor abnormalities are defined as defects that have unusual morphologic features without serious medical implications or untoward cosmetic appearance. Most minor abnormalities involve the face, ears, hands or feet, and are readily recognized even on cursory examination (Holmes & Hassanein, 1988). The presence of three or more minor abnormalities in newborns is correlated with a 90 percent frequency of coexistent major abnormalities (Marden, Smith & McDonald, 1964), suggesting close association with morphogenesis in utero.
Thus, minor abnormalities may provide clues to developmental problems of possible prenatal origin. Evaluation of the head, face, eyes, ears and mouth must include general assessment of visual acuity and hearing. Examination of the chest, heart, spine, abdomen, genitalia, extremities, muscles and neurologic reflexes can reveal abnormalities that may be associated with retardation.

EPIDEMIOLOGY

Over the past 50 years the prevalence and incidence of mental retardation have been affected by changes in the definition of mental retardation, improvements in medical care and technology, societal attitudes regarding the acceptance and treatment of an individual with mental retardation, and the expansion of educational services to children with disabilities from birth through age 21. The theoretical approach to determining the prevalence of mental retardation uses the normal bell curve to estimate the number of individuals whose IQ falls below the established criterion score. For example, 2.3% of the population of the United States has an IQ score below 70, and 5.5% has an IQ score below 75. However, this estimate does not account for adaptive behaviour skills. Based on empirical sampling, Baroff (1991) suggested that only 0.9% of the population can be assumed to have mental retardation, yet the proportion of cases that can be attributed to a known cause is estimated to be only 30% to 50% (Curry et al., 1997). An understanding of the epidemiology of MR of unknown cause may lead to the identification of characteristics that might be direct causal factors or lie somewhere along the causal pathway (Croen, Grether & Selvin, 2001).

Following a review of the most recent epidemiological studies, McLaren and Bryson (1987) reported that the prevalence of mental retardation was approximately 1.25% based on total population screening. When school age children are the source of prevalence statistics, individual states report rates from 0.3% to 2.5% depending on the criteria used to determine eligibility for special educational services, the labels assigned during the eligibility process (e.g., developmental delay, learning disability, autism, and/or mental retardation), and the environmental and economic conditions within the state (U.S. Department of Education, 1994). It is estimated that approximately 89% of these children have mild mental retardation, 7% have moderate mental retardation, and 4% have severe to profound mental retardation. In addition, McLaren and Bryson (1987) report that the prevalence of mental retardation appears to increase with age up to about the age of 20, with significantly more males than females identified.

CONCLUSIONS

Mental retardation is a genetic disorder manifested in significantly below average overall intellectual functioning and deficits in adaptive behaviour. A number of environmental, genetic or multiple factors can cause mental retardation. In at least 30 to 50 percent of cases, physicians are unable to determine etiology despite thorough evaluation. The systems review of the child should be complete, with special attention to growth problems, history of seizures, lethargy and episodic vomiting. The systems review of the child should be complete, with special attention to growth problems, history of seizures, lethargy and episodic vomiting.

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