Pediatric Neurology Part I: Chapter 21. Developmental abnormalities and mental retardation: diagnostic strategy (Handbook of Clinical Neurology)

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Intellectual disability formerly called mental retardation (MR) is defined as having an IQ score below 70; the term “developmental delay” (DD) is preferred for young children. A detailed clinical history including a three-generation pedigreee and physical examination are the fundamental steps in achieving an etiological diagnosis in MR. Physical examination should be performed with special emphasis on dysmorphological and neurological exam. Genetic studies have priority in the laboratory investigation of a child with MR. Routine karyotyping is recommended regardless of the degree of MR. Fragile X studies are strongly recommended in both females and males with unexplained MR, especially in patients with a positive family history and typical physical and behavioral features. FISH analysis of subtelomeric regions should be reserved for selected patients. Inborn errors of metabolism are seldom seen as the causes of isolated MR but should be considered in the differential diagnosis of patients with MR/DD in populations where the rate of consanguineous marriages is high. Neuroimaging studies should be performed on an indication basis such as abnormal brain size or neurological findings. It is essential to diagnose the underlying etiology of MR for recognition of treatable disorders, determining prognosis, family counseling, and providing prenatal diagnosis when possible.

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