DEVELOPMENTAL DISORDERS

VALUE OF CLINICAL EXAMINATION IN PREDICTING ETIOLOGY OF GLOBAL DEVELOPMENTAL DELAY

The clinical features at initial examination that proved of value in predicting the etiology of global developmental delay (GDD) were analyzed in a retrospective study of 261 unselected young children seen in an academic pediatric neurology practice, 1994-2004, at Montreal Children's Hospital, Quebec, Canada. Mean age at initial evaluation was 2.8 +/- 1.3 years. The male/female ratio was 2.84:1. GDD was mild in 12.2% patients, moderate in 43%, and severe in 44.1%. Etiologies defined in 98 (38%) patients included: genetic syndrome/chromosomal abnormality in 24 (24%), intrapartum asphxia 22%, cerebral dysgenesis 16%, psychosocial deprivation 11%, and toxin (eg alcohol) exposure 7%. Etiologies were prenatal in origin in 63%, perinatal in 22%, and postnatal in 15%. Clinical features predictive of eventual etiological diagnosis were: female gender, abnormal prenatal/perinatal history, the absence of autistic symptoms, microcephaly, abnormal neurologic examination, and dysmorphic features. In 113 children without abnormal clinical features, screening tests (karyotype, fragile X genotyping, and neuroimaging) provided the etiology in 18 patients. (Srour M, Mazer B, Shevell MI. Analysis of clinical features predicting etiologic yield in the assessment of global developmental delay. **Pediatrics** July 2006;118:139-145). (Respond: Michael Shevell MD CM, Room-A514, Montreal Children's Hospital, 2300 Tupper Street, Montreal, Quebec, Canada H3H 1P3).

COMMENT. The etiological yield based on clinical features in an unselected series of children with GDD is 38%, and 55% in the absence of autistic symptoms. Children with GDD and autistic features are less likely to have an etiology identified. Screening tests may supplement the yield with diagnoses defined in an additional 18%, when clinical findings are not helpful. The value of neuroimaging in the evaluation of global developmental delay demonstrated in this study supports the recommendation advanced by the Quality Standards Subcommittee of the AAN and Practice Committee of the CNS (Shevell M et al. **Neurology** 2003;60:367-380). In the absence of specific clinical features, a stepwise approach is recommended: first an MRI (nonenhanced), followed by cytogenetic screening, including fragile X, metabolic tests, and genetic consultation. (see **Ped Neur Briefs** February 2003;17:13-14). A review of the literature found that the MRI had the highest diagnostic yield (55%), and metabolic screening the lowest (1%). State-based newborn screening programs will identify some metabolic disorders shortly after birth (phenylketonuria, congenital hypothyroidism, sickle cell disease, and galactosemia). All children with GDD should have auditory and visual screening tests, and in 32 states a newborn hearing test is a requirement.

MRI in developmentally delayed and autistic children. An MRI study of 45 children examined by child psychiatrists (mean age 43 months; 41 male), 20 with autism, 12 with PDD, 4 mental retardation, and 9 language delay, found 22 (49%) with abnormalities. Four (8.5%) had arachnoid cysts, 3 located in the middle cranial fossa; 1 had Chiari I malformation, and 3 had dilated Virchow-Robin spaces, all nonspecific for GDD or autism. (Zeegers M et al. **Brain Dev** Sept 2006;28:495-499). (E-mail: m.zeegers@umcutrecht.nl).