

RETT SYNDROME

RETT SYNDROME SYMPTOMATOLOGY IN ADULT MALES

A survey of 297 institutionalized adults with mental retardation to determine the prevalence of symptoms of Rett syndrome is reported from the Medical Center Rehabilitation Hospital, Grand Forks, ND and University of North Dakota School of Medicine. The survey suggests that a) male and female adults as a group express the individual symptoms of Rett syndrome with equal frequency, b) as individuals, only females appear to exhibit the full criteria for Rett syndrome, c) males do exhibit particular symptoms of the syndrome, and d) no individual symptom or two symptoms separate males and females. The prevalence rate for Rett syndrome in the institutionalized population of persons with mental retardation was 1:46 for females and 1:116 for both sexes. Midline hand stereotypies, hyperventilation, impaired ambulation, seizures, and absence of prenatal complications all occurred with equal frequency in both sexes. (Burd L et al. Rett syndrome symptomatology of institutionalized adults with mental retardation: Comparison of males and females. AJMR March 1991; 95:596-601).

COMMENT. The oldest of the four females who met the strict and necessary criteria for the diagnosis of Rett syndrome was 40 years of age. The female sex is no longer considered a necessary diagnostic criterion and a less restrictive symptom complex is proposed by some authorities.

RETT SYNDROME AND MITOCHONDRIAL ENZYME DEFICIENCIES

Three children with Rett syndrome and normal muscle mitochondrial structure but abnormalities in mitochondrial respiratory chain enzymes are reported from the Departments of Neurology and Pediatrics, Loyola University, Stritch School of Medicine, Maywood, IL. The children, all girls, were three, four, and 13 years of age. The 13 year old had normal development until 15 months of age when she stopped talking and developed an abnormal gait and loss of hand use. At six years of age, self-injurious behavior and head hitting developed and at age nine years, she had atonic and clonic seizures. Her behavior was autistic and hyperactive. Hand wringing and mouthing were constant while awake. The head circumference was 49 cm (below the 2nd percentile). There were low levels of cytochrome c-oxidase and succinate cytochrome c reductase in all cases. Muscle morphology by light and electron microscopy was normal. (Coker SB, Melnyk AR. Rett syndrome and mitochondrial enzyme deficiencies. J Child Neurol April 1991; 6:164-166).

COMMENT. Mitochondrial alterations in muscle, including distention, vacuolation, and membranous changes, have been described in children with Rett syndrome. (See Ped Neur Briefs Sept 1989).