



Pediatric Neurology Part I: Chapter 31. Genetically determined encephalopathy: Rett syndrome (Handbook of Clinical Neurology)

Nadia Bahi-Buisson

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Rett syndrome (RTT) is a severe neurodevelopmental disorder primarily affecting females that has an incidence of 1:10000 female births, one of the most common genetic causes of severe mental retardation in females. Development is apparently normal for the first 6–18 months until fine and gross motor skills and social interaction are lost, and stereotypic hand movements develop. Progression and severity of the classical form of RTT are most variable, and there are a number of atypical variants, including congenital, early onset seizure, preserved speech variant, and “forme fruste.” Mutations in the X-linked gene methyl-CpG-binding protein 2 (MECP2) involve most of the classical RTT patients. Mutations in cyclin-dependent kinase like 5 (CDKL5) and FoxG1 genes have been identified in the early onset seizure and the congenital variants respectively. Management of RTT is mainly symptomatic and individualized. It focuses on optimizing each patient's abilities. A dynamic multidisciplinary approach is most effective, with specific attention given to epileptic and nonepileptic paroxysmal events, as well as scoliosis, osteoporosis, and the development of spasticity, which can have a major impact on mobility, and to the development of effective communication strategies for these severely disabled individuals.

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