Rett syndrome: a review.

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Rett syndrome, first described in 1966 by Andrea Rett, is a neurodegenerative disorder of unknown aetiology, even if the syndrome is associated to increased cell-packing density, neurotransmitter metabolic alterations and genetic and developmental factors. This disability is found almost esclusively in females, nevertheless in literature male types of Rett variants are described. The prevalence of the syndrome has been estimated to be approximately 1/10000 girls.

The disorder is characterized by a loss of cognitive and motor retardation, impaired expressive and receptive language, decelerating head growth, loss of purposeful use of the hands, jerky truncal ataxia, social regression and stereotyped hand movements.

In Rett syndrome, it has been applied a system based on 4 clinical stages.

The present article presents a review of current knowledge in Rett syndrome, concerning diagnostic criteria, clinical characteristics of the 4 stages and management. Information is provided on the differential diagnosis, in particular with autism, cognitive and adaptive profiles.

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