

Rett Syndrome

Living with Rett syndrome is kind of like being on a roller coaster that never stops. It has it's ups and downs, the smooth but bumpy course, the twists and turns that somehow seem to come when you least expect them, and knock you back into the reality that your child is different from others, but that doesn't mean she can't learn and can't lead a full and impact-filled life.

Living with Rett Syndrome, Connie Coughlin, mother whose daughter has Rett/Rhett syndrome, taken from <http://members.tripod.com>

Rett syndrome affects females almost exclusively and is a neurodevelopmental condition characterised by 'normal' early development followed by a period of regression where skills are lost. As with any syndrome there is huge variation in those with Rett syndrome and deterioration, in varying degrees, may be noticed in expressive language use, gait, general learning ability, purposeful use of hands, distinctive head movements and head growth.

Rett syndrome is classified by the Diagnostic and Statistical Manual of Mental Disorders, (DSM-IV) (4th edn) as a developmental disorder of childhood. The symptoms are generally described in terms of four stages:

Stage 1: Early Onset (6–18 months of age): the slowing of development is generally subtle at this stage and consequently symptoms are often overlooked.

Stage 2: Rapid Deterioration/Destruction (1–4 years of age): this stage may be gradual or rapid. Typically, purposeful hand use and spoken language are lost at this stage. Gait patterns are laboured and unsteady, and periods of trembling or shaking may be evident. Slowing of head growth may be noticeable. Behaviours such as hand-wringing, hand-clapping or grinding of teeth may present.

Stage 3: Plateau/Pseudo-stationary (2–10 years of age): apraxia (difficulties controlling voluntary muscular movements in spite of the fact that there is no actual muscle weakness), motor difficulties and seizures may present at this stage. Alertness, attention span and communication skills may improve as may a general interest in the individual's surroundings.

Stage 4: Late Deterioration of Motor Skills (typically 10 years onwards): this stage can last for a number of decades and is characterised by reduced mobility, muscle weakness, rigidity, spasticity and scoliosis. The individual may lose the ability to walk (if he/she managed to acquire this skill) at this stage. Repetitive hand movements and seizures may decrease and eye gaze may improve. It is important to note that there is no cognitive loss at this stage.

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