

Rett Syndrome

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What is Rett Syndrome?

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Definition:

Neurodevelopmental Disorder that affects girls almost exclusively. Individuals grow normal in early growth (6-18 months) then slowed development, lose of purposeful hand movement, slowed brain and head growth, difficulty walking, seizures, and intellectual disability.

Common Characteristics:

- Repeated hand wringing, washing, or clapping motion
- Small head size (microcephaly)
- Severe problems with language, communication, learning, coordination, and other brain functions

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Other Symptoms:

- Loss of muscle tone (hypotonia)
- Breathing abnormalities
- Spitting or drooling
- Unusual eye movements - intense staring or excessive blinking
- Cold hands and feet
- Irritable
- Sleep disturbances
- Seizures
- Abnormal side-to-side curvature of the spine (scoliosis)

Life Expectancy:

Beyond the age of 40 years old

Population Statistics:

- In 99%+ there is no history of the disorder in their family
- 1 in 9,000 - 10,000 females are affected

Cause

- Mutation of MECP2 gene (usually)
 - MECP2 regulates activity of genes
- Abnormal amounts of protein are produced to cause other genes to be abnormally expressed

Rett Syndrome in the Media

- Most recent public research was published around 2016
- Rettsyndrome.org advocate and provide a community for those diagnosed with Rett Syndrome.

Treatment Options

- There is no cure
- Can do treatment for symptoms - manage
 - Medication - breathing irregularities, motor difficulty, control seizures
 - Occupational Therapy - Activities of daily living
 - Physical and Hydrotherapy - Mobility
 - Orthotics - braces, spints
 - Nutrition
 - Others depending on symptoms

Impact of Recreational Therapy

- Improving quality of life
- Providing adaptive leisure opportunities
- Assisting in increased or maintained mobility
- Working with a multidisciplinary team to meet client goals

Teaching Tips/Techniques

- Each individual may be very different with diagnosis
- Be patient, with intellectual disability or speech impairments it may take longer for a response
- Use modeling
- Assume that the individual understands you
- Give as much independence as possible
- Explain before participation (front loading)
- One task at a time
- Look for body language or eye movement for responses

Resources:

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International Rett Syndrome Foundation. (2019). Home. Retrieved July 22, 2019, from <https://www.rettsyndrome.org/>

Keith, J. (Ed.). (2016). *Autism and pervasive developmental disorders sourcebook*. Retrieved from <https://ebookcentral.proquest.com>

Rett syndrome - Genetics Home Reference - NIH. (2019, July 16). Retrieved July 22, 2019, from <https://ghr.nlm.nih.gov/condition/rett-syndrome>