Rett Syndrome

What is it?

Rett Syndrome is caused by a MECP2 gene mutation. It is a non-inherited, genetic, neurological disorder. It is most commonly diagnosed in girls because the disorder is associated with the extra X chromosome found in female babies. Boys who are born with Rett Syndrome do not usually survive past infancy.

Rett Syndrome (RTT) is a rare disorder that has been diagnosed in 1 in 10,000 female births, with less than 1,000 children diagnosed in the United States each year.. There is nothing that the mother did during her pregnancy to cause this disorder nor is it inherited from either parent. It is important to stress this, as so often parents look to their own medical history and/or behavior to place guilt.

What are the symptoms?

There are several behaviors that a child with RTT exhibit, however, if your infant is demonstrating some of these behaviors it is not a guarantee that it is because of Rett Syndrome.

- Repetitive hand wringing while awake
- Loss of purposeful use of hands
- Loss of speech
- Loss of mobility
- Breathing difficulties

If your child exhibits any or all of these symptoms, we strongly advise that you seek the advice of your Pediatrician. There is a blood test that can be administered that will determine if your child has RTT. According to RettSyndrome.org:

"Rett syndrome is confirmed with a blood test to identify the MECP2 mutation. However, since the MECP2 mutation is also seen in other disorders, a Rett syndrome (RTT) diagnosis requires either the presence of the MECP2 mutation or fulfillment of the diagnostic criteria or both."

There are several Genetic laboratories around the United States that offer extensive testing, research and clinical trials. Visit https://www.rettsyndrome.org/about-rett-syndrome/rett-syndrome-diagnosis/ for the specific locations.

When is RTT typically diagnosed?

Most children born with RTT begin to show symptoms between six and 18 months of age. It is during this time when infants begin to demonstrate movement and speech patterns; both of which are impacted by this disorder.

What is the prognosis?

There are different ranges of RTT; while some with the disorder will have the ability to walk, others may never become mobile. However, there is on-going research and medical assistance that can help your child develop the skills to manage some of their personal daily needs.

Most with RTT will need some assistance with mobility, eating, breathing and speech; needs that will increase as the child moves through the different stages of the disorder.

The medical association recognizes four states of RTT:

Stage I – Early Onset Stage
Age: 6 months to 1.5 years
Duration: Months

Stage II – Rapid Destructive Stage
Age: 1 to 4 years
Duration: Weeks to Months

Stage III – Plateau Stage
Age: Preschool to adulthood
Duration: Decades

Stage IV – Late Motor Deterioration Stage
Age: When ambulation is lost (those who never ambulate move from Stage II to IV)
Duration: Up to decades

At this time, there is no known cure for the disorder and patients diagnosed with RTT are rarely able to live independently.

How does it impact communication skills?

Loss of speech is one of the primary symptoms of RTT, however, that does not mean that a child with the disorder is unable to think, comprehend or communicate. The use of augmentative devices, like those offered through PRC, provides life-altering assistance for those with RTT. Our professionals work with you to determine the best device available for your child. Additionally, our devices are modified to meet your child's needs and extensive training is offered for both the care giver and the child.

Resources

Primary source of information, resources, research and support: RettSyndrome.org

Additional Resources:

- **US Department of Health and Human Services:** https://www.nichd.nih.gov/health/topics/rett/resources/patients
- **Claire's Crusade:** http://www.clairescrusade.org/resources/resources/?view=mobile
- **National Organization for Rare Disorders:** https://rarediseases.org/organizations/international-rett-syndrome-foundation/
- **Cleveland Clinic:** https://my.clevelandclinic.org/health/articles/6089-rett-syndrome/resources
- **Boston Children's Hospital:**
  http://www.childrenshospital.org/centers-and-services/programs/o-_-z/rett-syndrome-program/patient-resources

Support

Facebook Rett Syndrome Family Support Forum,
https://www.facebook.com/groups/250235058326930/

Girl Power 2 Cure: https://www.girlpower2cure.org/