What is Rett Syndrome?

Rett Syndrome (RS) is a rare neurological developmental disorder, afflicting roughly one in 10,000 people of all racial and ethnic groups. It is seen almost exclusively in females, although it can occur rarely in boys. RS is the most physically disabling of the family of the autism spectrum disorders. Individuals are born healthy but then the culprit gene mutates, destroying speech and normal movement. A classic sign is hand movements such as clasping or wringing. What goes awry: a gene called MECP2 that is supposed to switch off other genes involved in the maturation of neurons. When MECP2 shuts off in RS, the brain cells don't die, but they don't keep developing. The identification of the gene in late 1999 in the lab of Dr. Huda Zoghbi at Baylor College of Medicine, after a 15- year search, was a crucial breakthrough that has catapulted the RS research field into the scientific limelight. Today, there are hundreds of laboratories around the world studying RS.

History

Dr. Andreas Rett first recognized RS in his Vienna clinic in 1963. He found another six similar patients and by 1966 was able to report on studies of 22 girls.
However, it was not until the disorder was reported in the english language by Dr. Bengt Hagberg of Sweden in 1983, that RS was internationally recognized.

Types of Rett Syndrome

•Classic RS: those who meet the consensus diagnostic criteria.
•Atypical RS: those who do not meet all of the diagnostic criteria for classical RS. The diagnosis of atypical RS must include at least three of the primary criteria and five of the supportive criteria. Atypical RS accounts for 15 to 20% of all RS diagnoses.

Diagnostic Criteria

•Period of apparent normal development until 6-18 months
•Purposeful hand use is replaced by stereotypical hand movements
•Loss of verbal language
•If able to walk, the gait is usually wide- based and stiff legged
•Normal head circumference at birth followed by slowing of the rate of head growth (there is a subset of girls whose rate of head growth does not decelerate)

Supportive Criteria

- •More than 95% will have a MECP2 gene mutation
- •Breathing pattern irregularities which include hyperventilation, breath holding, apnea, air swallowing
- •EEG abnormalities
- •Seizures
- •Scoliosis
- •Teeth grinding
- •Gastrointestinal issues
- •Biting/Chewing/Swallowing difficulties
- •Poor circulation to legs and feet
- •Decreased mobility with age
- •Muscle rigidity/spasticity/joint contractures
- •Abnormal sleep patterns
- •Irritability and agitation

Types of atypical RS

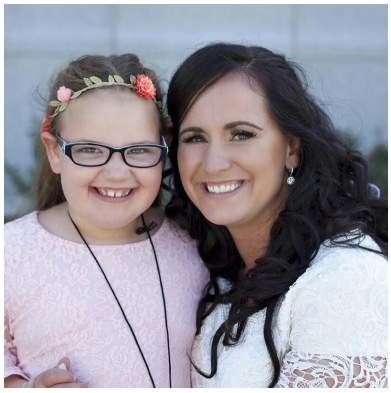
1.Congenital Onset RS: developmental delay is noticed shortly after birth with no early normal development; or severe seizures in early infancy impairing early development.
2.Late Onset RS: signs are delayed beyond the typical 18 month onset, in some cases to age 10 years or more.
3.Preserved Speech RS: milder features are seen.
4.Male RS: May be seen in males with Klinefelter (XXY) or somatic mosaicism.

What can be done?

At the present time, there is no cure for RS.
Treatment for the disorder focuses on the management of symptoms.
A team approach is essential to provide coordinated services including Neurology, Orthotics, Occupational, Physio and Communication therapy.
Special equipment such as braces to arrest scoliosis, splints to modify hand movements, ankle foot Orthotics to help with mobility and various daily living aides can be helpful.
At times, nutritional programs to maintain adequate weight are needed.

How you can help

Spread the word about RS by giving this pamphlet to your doctor, friends and family
Visit our website to learn more (www.rettsyndromealberta.org)
Become a Member of our Society
Make a Donation



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