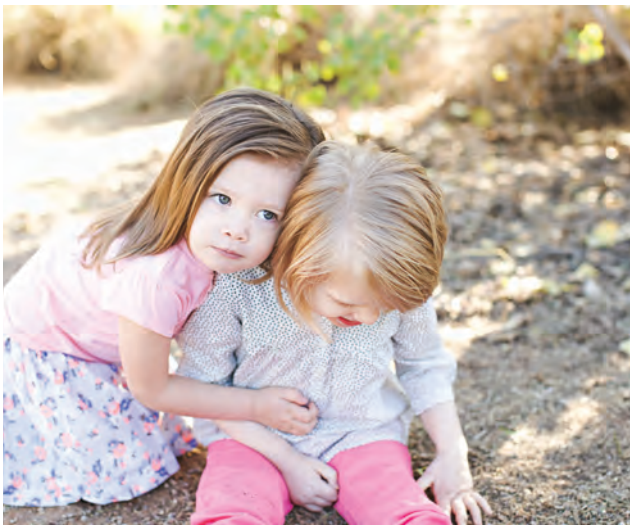
RETT UNIVERSITY

RETT SYNDROME - AN INTRODUCTORY GUIDE

Rett Syndrome



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Rett Syndrome is a neurological/movement disorder which predominantly affects girls (approximately 1 in 10,000). Many attain developmental milestones from birth to age two or three, but then begin to regress. Others are slow to develop from birth. It affects all aspects of motor control. In addition, there are cardiac, respiratory, gastrointestinal and orthopedic concerns. Rett is considered a "full-spectrum" disorder with varying severity and phenotypes depending on the child's unique genetic makeup, specific mutation, and/or percentage of cells skewed to use the mutated vs. the healthy copy of the Rett gene.

Most girls have repetitive hand wringing and/or mouthing, although the age of onset varies. Another common symptom is disordered breathing to include hyperventilating, breath holding and shallow breathing. Teeth grinding is also prevalent.

Other facts:

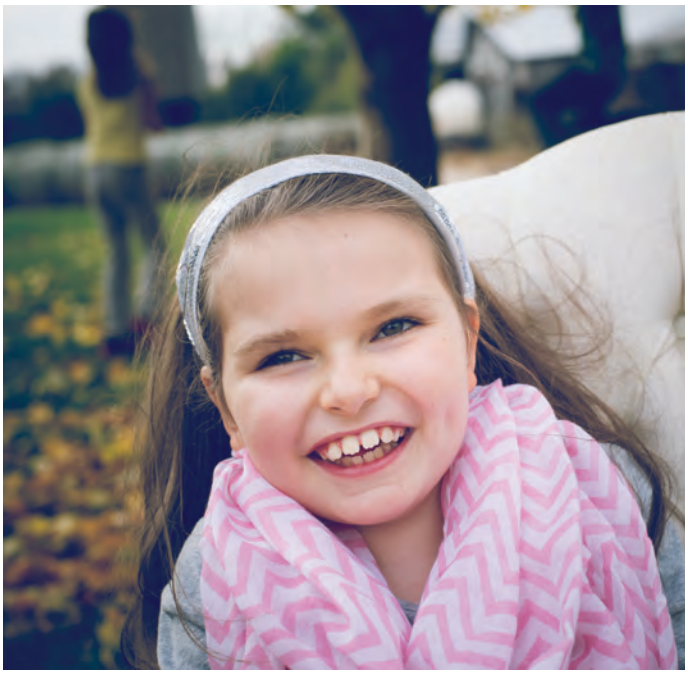
- Caused by a single gene mutation that leads to underproduction of an important brain protein.
- The leading genetic cause of severe impairment in girls – most cannot speak, walk or use their hands.
- Despite their physical disabilities, girls with Rett Syndrome are believed to be functioning mentally at a much higher level than previously thought.
- As prevalent as Cystic Fibrosis, ALS and Huntington's.
- Another little girl is born with Rett Syndrome every 90 minutes.
- Rett Syndrome is a potentially REVERSIBLE disorder. Research has proven once protein levels are back to normal levels, symptoms subside.

Today, there is no cure. But Rett Syndrome is CURABLE!
Amidst the mass confusion going on within their central nervous systems, they are smart, strong, and waiting bravely for us to unlock the door to their recovery.
Researchers at Harvard, Baylor, UVA, Weill Cornell, Emory, Salk Institute, University of Edinburgh and dozens of other top-notch institutions are delving into Rett Syndrome.

Why?

Not only will their work help thousands of girls and women worldwide, but their findings have a direct impact on research on Autism, schizophrenia, bipolar disorder, Alzheimer's and many other disorders.
In essence, our girls may hold the key to a cure for millions of people suffering from dozens of disabilities.

How is it diagnosed?



The diagnosis of Rett Syndrome will typically be given by a pediatric neurologist, clinical geneticist, or a developmental pediatrician.

A pediatrician or therapist familiar with Rett Syndrome may believe it to be the cause of the child's low muscle tone, scoliosis, seizure disorder, and/or loss of speech/mobility (among other symptoms), but will generally refer the patient to a specialist more familiar with neurological and genetic disorders. Once referred to a specialist, they may look at all the symptoms presented by the patient and suggest different tests to rule out other disorders or syndromes including, but not limited to, Rett Syndrome.

Many girls can be given a clinical diagnosis based on observations by one of the above specialists. A blood test can be preformed to confirm a gene mutations known to be a factor in Rett Syndrome, including the main gene MECP2. There are also individuals who have mutations in the CDLK5 and FOXG1 genes who are diagnosed with atypical or congenital Rett Syndrome

Understanding their needs

What skills/strengths do these students have?

- Rett patients are generally social and respond favorably to touch and interaction.
- They have a strong eye gaze which remains consistent despite motor issues which can be leveraged for communication and computer access.
- They are capable of learning both physically and academically.
- They are able to communicate basic emotions through body language and vocalizations.
- Their receptive learning/understanding level is high.
- Given a way to communicate they can indicate higher level reasoning and learning.
- Usually love books and the computer.
- Able to persevere past the apraxia in a supportive, encouraging environment.

What key things impede their ability to progress?

- Lack of stimulating environment (physical and academic)
- Severe apraxia
- People thinking it is more of a cognitive issue than an apraxic issue
- Poor balance
- Poor sensory integration
- Erratic breathing
- Disruptive, uncontrollable hand movements
- Extreme anxiety
- Seizure activity

What should I take into account when presenting tasks or trying to educate a Rett student?

- Assume competence cognitively
- Look for things that are highly motivating to incorporate into the task or into the "reward" for participating. (Food, music, favorite book)
- Expect anxiety when presented with new things. Keep first exposure short and then increase time at each subsequent session.
- Communicate with child what your expectations are.
- To lessen anxiety, have a way for the child to communicate with you.
- Read their communication first – be a "meaning maker" to all their attempts
- Give fun ways to communicate: real comments, expressions, arguing, etc. -- not just requesting.
- Give them choices.
- Be sure they are properly/securely positioned.
- It helps to move items you want them to attend to in order to recapture attention
- Do not necessarily take non-response or frustration as a sign the child cannot do the activity. Often, these girls are bored and unmotivated at the cognitive level being presented.
- Try not to "undershoot" the girls for example by putting them in repeated demand situations (look at ...., point to...)

What kind of pace is beneficial?

It is important to break down tasks into very small steps and stretch out over a long period of time with a lot of repetition. Each step should provide an opportunity for success.
Example: *Learning to use a light switch.*

- Stage One: Approach switch with child and verbally talk about the switch and turn it off for them.
- Stage Two: After verbal prompt, use hand-over-hand to model.
- Stage Three: Approach light, give verbal prompt, but wait for ANY reaction (head turn, hand jerk, etc) before using hand-over-hand.
- Stage Four: Follow above, but wait for increased reaction (hand touching anywhere on the wall).
- Stage Five: Follow above, but wait for increased more precise reaction (hand touching general vicinity of switch).

*It is important to give enough time and wait for initiation of response from child. Sometimes their bodies are not able to react for 30-60 seconds or more. If they look toward the item accept that as communication and build on it.

What is the best environment to work in?

- The ideal setting would be a quiet place with little distraction or sudden noise.
- Avoid bright lights or shiny objects which can be very distracting.
- Keep the surroundings predictable for each session.
- Incorporating a sensory rich environment can increase success (music, rhythm, deep pressure/weighted vest, etc)
- Incorporating a consistent mode of interactive communication for the activity

What do these students tend to enjoy?

Music, water, face-to-face contact, weight/pressure/massage, swinging, small group or one-on-one interaction with normal peers. People who are fun, funny, engaging, exciting, and those who wait for them and believe in them.

What generally causes stress in Rett students?

- Not being able to communicate
- Change / new things
- Sudden noise
- Physical pain from gas pains, air swallowing, constipation and other issues.
- People who don't talk to them like they are people
- Remember: Avoid negative comments/commentary in their presence – they understand and will internalize the information.

Communication

What kind of communication can I expect/work towards with a child with Rett?

Many of the Rett patients have had success with the following:

- Big Mac and Step-by-Step switches
- Yes/ No cards
- Boardmaker icons
- Eye-gaze computer systems

If they are not capable of "swatting" at a switch with their hand, consider other placements for access with their head or cheek.

- Look for every opportunity to assign meaning to their movements and vocalizations.
- Eye gaze systems (hi and lo tech) will be the most consistent over time.
- They are capable of symbol recognition as well as learning to spell/read.

Which key classroom/therapy supports may be needed?

- **Positioning:** chair with proper back and foot support with tray for feeding and activities, stander with activity tray
- **Communication:** Various switches, eye gaze system (low tech boards and hi tech infared computer systems are available)
- **Sensory:** Weighted vest, weights for wrists and ankles, swing, music player

Other:

- Arm braces to give hands a break from mouthing and wringing
- Small bite-size snacks and water should be available throughout the day
- Laminated materials
- Slant board for easier reading

Goals

What would be helpful in writing their IEP goals?
Progression in their learning follows the same patterns as "normal" children, it is the timeline that is extended to a great degree.

What is the long-term prognosis?
While many patients with Rett Syndrome live into their 40s and 50s, they require total, 24-hour-a-day care. Ongoing therapy is critical to their well-being.

What is the research outlook?
Rett Syndrome is most often caused by a sporadic mutation in the MECP2 gene, causing underproduction or misproduction of a protein that plays a key role in turning on and off other genes. In 2007, researchers were able to reverse the symptoms of Rett in fully mature mice only days from death by reestablishing normal protein levels in the brain. In 2012, researchers were able to replace the faulty gene in mice using gene therapy which led to a complete recovery. It is possible that Rett Syndrome will be the first curable childhood neurological disorder.

Personal notes