

Special acknowledgement to **Kathy Hunter** (<http://retthelp.org>), **Author of The Rett Syndrome Handbook I & II and founder of IRSA (International Rett Syndrome Association)** which merged and became the **International Rett Syndrome Foundation** (<http://rettsyndrome.org>) who's original online publication, What Kids Ask About Rett Syndrome, was in part the basis for this booklet which was been updated with information in part from the National Institute of Health Publication #01-4863 and I part from our personal experiences with our daughter.

Please visit <http://UnicornMeadows.org> to learn more about Rett Syndrome.

# What Kids (& Parents) ask about Rett Syndrome



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Dedicated to our beloved daughter  
Laura Amy Krivoshik \*



who was diagnosed with Rett Syndrome  
when she turned 9 in September 2002.

\* Laura has atypical Rett Syndrome (genetically confirmed) and talks a lot but has trouble communicating.

or to help her motor coordination and breathing. She may need special assistance in eating.

She also needs special therapies like:

- Physical therapy to help her keep flexible and able to move and walk better;
- Occupational therapy to help her use her hands for eating and drinking, brushing her teeth, and even playing with toys or games;
- Speech therapy to teach her other ways to communicate because apraxia makes it very difficult to talk;
- Music therapy gives her another enjoyable way to communicate and to make choices;
- Hydrotherapy, helps her move more easily by using warm water;
- Horseback therapy, helps strengthen her muscles, improve her balance, and lets her feel in control.

### Just like you

Even girls with Rett Syndrome are different from each other in some ways. A very few girls can talk, but have difficulty communicating and expressing themselves. Many of our girls go to special schools while some attend their local schools with special help. They learn in different ways, but they love to learn, just like you. They also love music, sports, animals, food and friendly people, just like you!

**So now you know about Rett Syndrome!**

regular kid. And all kids have the same smile! Get to know her and you will see.

## How do you get Rett syndrome?

You can't catch Rett Syndrome! It is caused by a fault in a gene she is born with. The fault almost always occurs spontaneously, it is not inherited. Genes are directions in all of our body cells that tell our body's cells how to make the substances that we need to function well. Each cell has thousands of genes, and each gene has thousands of instructions (codes). They determine if we will have blue or brown eyes or will be short or tall and all of the other characteristics that make us all different. The codes are long and complicated, so sometimes there are mistakes. Each of us has some of these mistakes (mutations) but they are harmless. Sometimes, the directions get so mixed they cause conditions like Rett Syndrome. Only in late 1999 was the gene responsible for Rett Syndrome identified and a test became possible.

## How come she seemed OK as a baby?

The mixed-up gene is there even before she was born. It just takes some time for enough problems to develop for us to notice that something is wrong.

## How is Rett Syndrome treated?

There is no cure at this time. Her symptoms can only be treated. She may need medicine to control her seizures

## Why have I never heard about Rett Syndrome?

Like most people you probably have never heard of Rett Syndrome, because it is quite rare affecting only one in every 10,000 to 15,000 girls. But you might know or have met a girl with Rett Syndrome! She may live in your neighborhood or attend your school. You probably noticed right away that she makes special movements with her hands almost all the time. Do you wonder why? It's because Rett Syndrome affects how her brain works.

## Why is it called Rett Syndrome?

Rett Syndrome was named for Dr. Andreas Rett, who first identified the syndrome. A syndrome is a group of symptoms - which together identify a special condition. Girls with Rett Syndrome have the same symptoms, although to different degrees. Dr. Andreas Rett wrote the first article about Rett Syndrome in 1966, but it was not until 1983 that an article was published in English.

## What are the symptoms of Rett Syndrome?

Rett Syndrome almost always happens in girls. She seems to be healthy and normal when she is born. She smiles, sits up and may begin to walk and talk. Within the next couple of years, though, she begins to lose her skills and is no longer able to do what she once could.

Here is a list of typical symptoms for Rett Syndrome:

- She begins to make different hand movements such as clapping, "washing her hands" and putting her hands in her mouth;
- She loses the ability to use her hands to pick up and hold things;
- She can no longer say words;
- If she can walk, her legs are stiff and wide apart. She might walk on her toes;
- When upset or scared, her body trembles or shakes;
- Her head does not grow at the rate it should.

She may also have other symptoms like:

- She may have seizures, which happen when her brain creates extra powerful jolts (seizures can be very scary to watch, but they do not hurt her);
- Her feet are very small and cold.
- She is small for her age.
- She may have trouble chewing or swallowing.
- She may breathe differently, either holding her breath, called "apnea", or huffing and puffing, called "hyperventilation".
- She may have scoliosis, where her back curves so she is leaning to one side or leaning forward.
- She may grind her teeth.
- She is almost always thin, even though she eats a lot.
- She may be irritable and not sleep well.
- As she gets older, she may not be able to move and walk as well as she did when she was younger.

## What kind of problems does a girl with Rett Syndrome have?

She has a lot of problems that affect her in many different ways, but the biggest problem is "apraxia" where her body cannot do what her brain tells it to - even simple things like moving and talking. Just imagine how hard it would be to know what you want to do and not be able to do it, or to understand what someone says but not be able to show that you understand. It can be very frustrating having Rett Syndrome.

## How much does she understand?

Most Rett Syndrome girls are not able to talk but that doesn't mean she has nothing to say! While she probably understands everything you say, she cannot speak or react the way you expect or want her to. Pay attention to her body language and her eyes - they tell you a lot! When she is irritable or loud, she is probably trying to tell you something important in the only way she can. Be patient, she needs extra time to respond and react.

## How can I help her?

Talk to her like you would someone else her age, but give her extra time to respond. Sometimes it takes several minutes before she can give an answer or make a movement to show you she understands. Include her in everyday activities even if she can't participate fully. She is really more like you than she is different - just a