N I C H D

National Institute of Child Health and Human Development

National Institutes of Health

Autism Research at the NICHD

For more information, contact the NICHD at:

NICHD Clearinghouse
P.O. Box 3006, Rockville, MD 20847
1-800-370-2943
Fax: 301-984-1473
E-mail: NICHDclearinghouse@mail.nih.gov
Internet: www.nichd.nih.gov/autism

Rett Syndrome

In October 1999, scientists sponsored by the National Institute of Child Health and Human Development (NICHD) made a remarkable announcement—they discovered that a change in the sequence of a single gene can cause Rett syndrome. Rett syndrome is one of many conditions classified as an autism spectrum disorder, which means it is not autism but has features that are similar to autism. This disorder causes autism-like symptoms, such as poor language skills, repeated hand motions, and decreased social contact in girls. These symptoms begin sometime between ages six months and 18 months, after apparently normal development.

With this discovery, NICHD researchers have their first glimpse into this baffling disease. These researchers join parents, families, and communities affected by the disorder in hoping that the discovery will lead to better diagnosis, treatment, and maybe even prevention of Rett syndrome.

What is Rett syndrome?

Imagine you are the parent of a baby girl. When she is born, the doctor tells you she is normal and healthy. You watch her start to become a little person. She smiles at you and your family when she's six weeks old. She picks things up with her thumb and first finger when she is seven months old. At 10 months, she is rolling and crawling her way into everything. You take pictures of her first birthday, where she is sitting up without your help and smiling at the camera. She can even say her own version of "cheese."

Now imagine that same daughter at age two. She can no longer sit up and doesn't grasp with her fingers. She starts having seizures. By the time she's three, she is always grinding her teeth and stops talking. When she turns six, her spine starts to curve, which limits how well she can move. She screams and laughs during the night for no reason, but doesn't respond to or interact with others. By her eighth birthday, she can't move on her own and can't talk.

The nightmare you just read is real for parents of girls with Rett syndrome. This tragic disorder causes some girls, whose growth, language skills, and personalities seemed normal before, to stop developing.

Sometime between their sixth and 18th month of life, these girls' development actually goes backward. They stop talking. They can't control their feet when they walk. They stop using their hands to do things, or start wringing their hands all the time. These girls stop responding to their parents and pull away from social contact with others.

Rett syndrome is a challenging disease for most of the families who are touched by it. Although many girls with the disorder live into their 40s, their lives are often not easy. Many of them can't walk or talk, but have to communicate with their eyes. They need special education, diets, and treatments for their various problems. Most girls with Rett syndrome can't care for themselves and need someone to care for them all of their lives.

What happens to girls with Rett syndrome as they get older?

In some girls with Rett syndrome, the body and mind keep growing and developing, but at a much slower rate. They have coordination problems, so they may not be able to walk backward or walk up the stairs. They also have learning disabilities, including problems remembering facts, understanding ideas, and solving problems. The lives of these girls are similar to the lives of people with other developmental disabilities, such as autism or Down syndrome.

Other girls with Rett syndrome lose more of their motor skills. They stop being able to sit up or use their hands. Some of them have seizures; others have trouble breathing while they are awake. Still others laugh or scream during the night for no apparent reason. A number of these girls develop scoliosis (pronounced sko-lee-oh-siss), which is a curving of the spine. By the time they reach their 20s, many of these girls are left completely helpless. They can't move and can't speak. These girls are at greater risk for dying suddenly and from unexplained causes.

What causes Rett syndrome?

As mentioned earlier, Rett syndrome is caused by a change in a single gene. Because this condition is relatively rare, affecting one female out of 10,000 to 15,000, researchers have long felt it probably involved genes.

What are genes?

Genes are very small pieces of hereditary material, which means that parents pass them on to their children. Every person gets half their genes from their mother and half from their father.

The pattern, or sequence, of your genes is like a blueprint that tells your body how to build its different parts. Your gene sequence controls how tall you are, what color your hair and eyes are, and other features of your body and mind. Changes in that blueprint can cause changes in how your body or mind develops.

Genes are found on chromosomes. Almost every cell in your body contains 23 pairs of chromosomes, 46 in all. Genes and chromosomes give the body all the information it needs to "build" a person. Of your 46 chromosomes, 44 help make your body and two control whether you're a female or a male. Females have two X chromosomes, and males have one X chromosome and one Y chromosome. Because Rett syndrome occurs only in girls, and girls have only X chromosomes, doctors decided to focus their research on the X chromosome.

How do genes cause Rett syndrome?

Scientists found that girls with Rett syndrome have a change in the pattern of one of their genes, specifically the gene that makes a protein called methyl cytosine binding protein 2 or MECP2. Normally, girls use the genes on only one of their X chromosomes; the genes on the other X chromosome are "switched off" by a complex set of chemical reactions in the body. MECP2 is the starting point of the process that "switches off" certain genes at certain times. Without it, these other genes aren't switched off.

In Rett syndrome, the body keeps making these materials, in large amounts, even when they are no longer needed. After several months, large amounts of these materials actually start to hurt the nervous system, instead of helping it to grow. This is why girls with Rett syndrome seem to grow normally until they are between six and 18 months old, but then stop developing and eventually lose developmental ground.

Because they have only one X chromosome, boys with Rett syndrome have only the changed gene for MECP2. Since they lack the "backup" or unchanged copy of the gene that girls have on their second X chromosome, boys with Rett syndrome die before birth.

Is there any cure or treatment for Rett syndrome?

There is currently no cure for Rett syndrome. However, girls with the condition can be treated for some of the problems associated with Rett syndrome. For example, physical and occupational therapists can help these girls overcome problems of coordination and movement, while speech therapists can help these girls learn to talk or communicate. There are also a number of medicines that can help prevent seizures and breathing problems that many girls with Rett syndrome experience.

How does the information about MECP2 affect girls with Rett syndrome?

Because Doctors know that MECP2 is missing in girls with Rett syndrome, and what MECP2 does in the body, they can explore ways to correct the problem. For instance, doctors might find a way to switch off genes that doesn't rely on MECP2. If doctors can slow or stop the progress of Rett syndrome, they may also be able to reverse its effects. This new information could also lead to ways to screen for Rett syndrome—to detect it before the girls start to feel the effects. In this way, doctors could start treating the girls much earlier, which could improve the lives of these girls.

The NICHD continues its efforts to understand Rett syndrome, in hopes of learning to slow, stop, and reverse its effects. The researchers involved, from both the Howard Hughes Medical Institute at Baylor College of Medicine and Stanford University, feel that the new information about MECP2 is a big step forward.

This new information also gives some insight into autism spectrum disorders, the group of conditions with similar symptoms that includes Rett syndrome. With an understanding of how these disorders affect the body, doctors will be better able to treat them. This knowledge is important not just for those affected by Rett syndrome, but also for any person touched by a developmental disorder.

For more information about the Rett syndrome research at the NICHD, contact the NICHD Clearinghouse at:

Mail: PO Box 3006, Rockville, MD 20847
Phone: 1-800-370-2943
Fax: 301-984-1473
Email: NICHDClearinghouse@mail.nih.gov

You can also learn more about NICHD research on Rett syndrome and autism spectrum disorders by accessing the NICHD Autism Web Page, at www.nichd.nih.gov/autism.

U.S. Department of Health and Human Services
Public Health Service
National Institutes of Health
National Institute of Child Health and Human Development

NIH Pub. No. 01-4960
June 2001