

## Have You Just Received Your Rett Syndrome Diagnosis?

We're here to help! IRSF has set up a Regional Representative Program to provide emotional support, information and resources to families affected by Rett syndrome. If you're in the United States, please contact the Rep in your state TODAY! If you are not located in the US, please contact the organization closest to you.

### Getting the Diagnosis of Rett Syndrome is Difficult

However, there are many people who are willing to help you, beginning with IRSF. Here are a few places to start:

- Visit our website often to learn the most current, accurate information about Rett syndrome
- Join an IRSF online community in order to build relationships and a support network with other parents just like you.
- Join the IRSF Partner program to stay connected with the Rett syndrome community through:
  - Newsletters; e-news alerts; discounts on Foundation merchandise, materials, conferences and education programs; special updates and announcements about research and self-help and education programs; and invitations to local clinics, events and program activities.
  - Take the time you need to grieve. Feeling depressed and overwhelmed is very natural. Find a grief counselor who is familiar with special needs families.
  - Make sure your child's medical team is knowledgeable about Rett syndrome. IRSF can help provide information and can refer you to the nearest specialist. Find out what educational services are available for your child. Call the special education department of your local school system.
  - Seek out information about financial assistance programs. Call the public health department, social work department of a local children's hospital, your Developmental Disabilities Administration, or local ARC.
  - Take care of yourself so that you will be able to better care for your child.
  - Don't try to plan for a lifetime in one day.
  - Remember that you are not alone.

You may have questions. Here are some answers from the Rett Syndrome Handbook:

### WHAT IS RETT SYNDROME?

Rett syndrome (RTT) is a unique developmental disorder that is first recognized in infancy and seen almost always in girls. It is found in all racial and ethnic groups throughout the world, and in every socioeconomic class.

## SHE SEEMED TO DEVELOP SO NORMALLY. WHAT HAPPENED?

RTT results from a chain of events beginning with a genetic mutation (change in a specific piece of DNA), which occurs at the time of conception.

The name of the mutated gene is MECP2 (methyl CpG binding protein 2), and it is pronounced "meck-pea-two)." It is always found at the end of the long arm of the X chromosome.

## WHAT DOES THE MECP2 MUTATION DO?

The MECP2 gene is a "housekeeping gene," responsible for telling downstream genes when to shut off. Scientists believe that lack of a properly functioning MECP2 gene allows other genes to come on or stay on at inappropriate times, disturbing the precisely regulated pattern necessary for proper development of the central nervous system. Development appears to be normal in early infancy until the time when the disrupted and normal brain development is altered. Without any of these specific factors, or with too much of any of them, selected regions of the brain remain developmentally immature, or are flooded with an overproduction of proteins and enzymes that are toxic to the central nervous system. The child appears to be developing normally in the first months of life because her early jobs do not demand much work from these factors until later in development. We now believe that the type of mutation and the number of cells impacted determine the stage when normal development will cease.

## DID I DO SOMETHING TO CAUSE THE MUTATION?

We do not know why this mutation occurs. Mutations occur naturally in everyone all the time and most do not cause problems. In fact, some mutations can result in improvements, like a better protein for using oxygen. Unfortunately, a mutation on the MECP2 gene results in RTT. Studies to date have not revealed any pattern in exposure to chemicals or radiation, or any correlation with demographics (where you live). The most likely explanation for the MECP2 mutation is that while the gene is forming and going through thousands of rapid duplications, it just stutters or burps, causing a change in the normal pattern of DNA. When this happens in much of the other of billions of pairs of DNA pieces, it just isn't noticeable.

## IS THE MECP2 MUTATION FOUND ONLY IN RETT SYNDROME?

We have recently learned that mutations in the same gene are involved in a number of other more well-known disorders, including autism, mental retardation, learning disorders, schizophrenia, and bipolar disorder.

## AT WHAT AGE DOES RETT SYNDROME BEGIN?

The age when RTT begins and the severity of different symptoms may vary. The child with RTT is usually born healthy and shows an early period of apparently normal or near normal development until six to eighteen months of life, when there is a slowing down or stagnation of skills. A period of regression then follows when she loses communication skills and purposeful use of her hands. Soon, stereotyped hand movements, gait disturbances, and slowing of the normal rate of head growth become apparent. Other problems may include seizures and disorganized breathing patterns while she is awake. There may be a period of isolation or withdrawal when she is irritable and cries inconsolably. Over time, motor problems may increase, while other symptoms may decrease or improve.

Answers adapted from the Rett Syndrome Handbook