

Where Can I Find More Information?

Rett Syndrome Association of Massachusetts, Inc. (RSAM)

217 South Street
Waltham, MA 02453
(781) 444-8164
(508) 757-9952
www.rettsyndromemass.org

The Rett Syndrome Program for Children and Women Children's Hospital, Boston

Department of Neurology
300 Longwood Avenue
Boston, MA 02215
(617) 355-8994
(617) 730-0279 (fax)
www.childrenshospital.org/rett

International Rett Syndrome Foundation (IRSF)

4600 Devitt Drive
Cincinnati, OH 45246
(513) 874-3020
www.rettsyndrome.org



**Help us find a cure
for Rett syndrome.**

RSAM Rett Syndrome Association of Massachusetts, Inc.

Rett Syndrome Association Of Massachusetts (RSAM)

The Rett Syndrome Association of Massachusetts was founded by parents of girls with Rett syndrome in 1989, at a time when little was known about this disorder. Originally created as a support group focused on education, the mission of RSAM as a 501 (c)(3) nonprofit today is to:

- Help families cope with the challenges of Rett syndrome in everyday life;
- Raise funds to promote biomedical research for treatments and a cure for Rett syndrome;
- Increase public awareness and knowledge among medical and educational communities.

Meetings are held throughout the year to help families access key resources and meet other families. Speaker forums are organized so that scientists and physicians can give research updates and address key medical issues. Educators and therapists provide important strategies that can be used to improve communication, mobility, and learning.



What is Rett Syndrome?

Rett syndrome is a developmental disorder seen primarily in girls in all racial and ethnic groups worldwide. The disorder was identified by Dr. Andreas Rett, an Austrian physician who first described it in a journal article in 1966. It was not until a second article about the disorder was published in 1983 that the disorder was recognized in the United States.

The course of Rett syndrome and the severity of symptoms varies from child to child. The child with Rett appears to grow and develop normally and then between six to 18 months of age, symptoms appear. Hypotonia (loss of muscle tone) is usually the first symptom. As the syndrome progresses, most



children lose purposeful use of their hands and the ability to speak. Other early symptoms may include problems crawling or walking and diminished eye contact. For many, the loss of functional use of the hands is followed by repetitive hand movements such as wringing, washing, or mouthing.



Other symptoms of Rett syndrome may include apraxia (the inability to perform motor functions), sleep problems, wide-based gait, teeth grinding and difficulty chewing, slowed growth, seizures, cognitive disabilities and breathing irregularities.

Rett syndrome affects about 1 in every 10,000 to 23,000 female births. Although rare, it is now seen in some boys. Rett syndrome leaves children dependent on others for all basic needs such as feeding, dressing, bathing, toileting, etc.



Cause

In 1999, a research team led by Dr. Huda Zoghbi of the Howard Hughes Medical Institute and Houston's Baylor College of Medicine, identified the cause of Rett syndrome as a defect in the MECP2 gene which is found on the X chromosome. Research has raised the possibility that this defective gene results not only in Rett syndrome, but in a wide range of disorders, including autism, schizophrenia, and learning disabilities.

Treatment and Outlook For Those with Rett Syndrome

There is no cure for Rett syndrome. Treatment for the disorder focuses on the management of symptoms. Medication may be needed for breathing irregularities and motor difficulties. Antiepileptic drugs are used to control seizures. Some children require gastrointestinal tubes for feeding. Others require equipment and aids such as braces to arrest scoliosis, splints to modify repetitive hand movements, and nutritional programs to help them maintain adequate weight. Physical and occupational therapies may prolong mobility, quality of life, and life expectancy.



Research

The recent discovery that mutations in the MECP2 gene cause Rett syndrome has promoted much interest in research among scientists. Many projects are focusing on understanding the role of this gene, its interaction with other genes, and whether Rett syndrome can be reversed. This research may also lead to a better understanding of other neurological disorders. Parents are hopeful that science discoveries will be translated into beneficial treatments for their daughters and that one day soon, a cure will be found.

How Can You Help?

Rett Syndrome Association of Massachusetts relies on donations to help support biological research and education. Please give as generously as you can to help us find a cure for Rett syndrome.

Enclosed is _____\$25 _____\$50 _____\$100 _____\$250 \$_____ Other

Name: _____

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Please make your tax deductible donations to:

Rett Syndrome Association of Massachusetts, Inc.
 217 South St.
 Waltham, MA 02453
 Our tax ID number is 04-3583648

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