

Background:

Andreas Rett, an Austrian pediatric neurologist, first described this unique neurological disorder in females in 1966. In 1983, Swedish neurologist Bengt Hagberg brought this condition to the attention of the international medical community.

Cause: X-linked genetic disorder:

Rett syndrome results from arrested development of the brain. It is not a degenerative disorder as once thought. Global prevalence rates range from 1 in 10,000 to 1 in 23,000 live female births. Initially scientists thought that Rett syndrome was lethal to males, but living males with the same genetic defect have now been identified. Researchers do not yet know the prevalence rate in males.

In the vast majority of families studied Rett occurs sporadically. However, in less than 0.4% of families, it may occur in more than one family member. In rare cases, mothers carry the gene for this mutation.

Until recently, Rett syndrome was a medical mystery. In October 1999, researchers traced the genetic basis for Rett syndrome to a defective gene called MECP-2 (meckpea-two) on the X chromosome. This mutation has been recognized in over 80% of children with classic Rett syndrome. Family studies contributed to the discovery of this gene.

Classic stages of Rett syndrome:

Although there is a wide variability in the clinical presentation, classic Rett syndrome follows a well recognized pattern. The type and onset of symptoms may differ from child to child. Some children lose functions early and others display potential to develop new skills as they get older.

Stage I (6-18 months)

- Most children have a normal birth history and early development.
- Most children have a mellow disposition.
- They tend towards “floppiness” in their body.
- Mild feeding difficulties may result from low muscle tone or loss of muscle control.
- Head growth may decelerate.
- A lack of developmental progress is noted and can be very subtle.

Stage II (1-4 years)

- Children experience either a gradual or sudden loss of acquired language, behavioral, social and psychomotor skills.
- There is loss of normal hand skills. Involuntary repetitive and movements may include: hands to mouth, hand washing, hand wringing, hand clapping, patting and touching. These movements cease during sleep.
- Shakiness of the trunk and tremors of the extremities are common, particularly when the child is excited or agitated.
- Breathing irregularities occur frequently including hyperventilation, breath holding and air swallowing with bloating.
- Children may grind their teeth or chew abnormally.
- Seizures may begin at any time.

- Vacant or staring spells may occur, but are not necessarily related to seizures.
- Many children have sleeping problems.
- Some learn to walk and have a wide-based stiff legged gait. Some individuals toe walk.
- Due to the similarities of the symptoms, doctors often mis-diagnose Rett syndrome as either Autism or Cerebral Palsy.

Stage III (Preschool and older)

- Symptoms appear to stabilize.
- Children interact and focus better.
- Stereotypical hand movements remain but may decrease in intensity.
- Sleep problems may continue.
- Muscle rigidity and dyspraxia (difficulty in planning motor movement) may interfere with movement.
- Swallowing problems can interfere with feeding.
- Growth slows, and decreased body fat and muscle mass may be observed.
- Constipation is a problem.
- Poor circulation in the extremities results in cold, blue hands and feet.
- Curvature of the spine and foot deformities may appear.
- Slow bone formation can result in unexplained fractures.
- Some show improved learning such as walking and increased use of hand function for basic skills such as finger feeding or using utensils.
- Some develop use of single words and understand directions.
- Puberty and fertility is unaffected and menstruation usually occurs at the expected age.

Stage IV (Any age)

- Individuals are unable to walk.
- Other symptoms continue.

Variations in Rett syndrome

Non-classic Rett syndrome occurs in approximately 1 in 45,000 and may explain the cause of developmental disabilities in many older women.

Early onset: These children have never shown normal development.

Early seizure onset: Although seizures are not necessarily the first manifestation of Rett syndrome, in some children this may be the first symptom.

Late onset: Some show functional loss in language and hand use. Hand stereotypes and other symptoms may appear later.

Preserved function: Some demonstrate the typical pattern of Rett syndrome but retain some language and hand use.

Rettoïd forms: Rett-like signs and symptoms may be seen in some children who have had neurological problems such as meningitis, encephalitis, tuberous sclerosis, or metabolic disorders. These should not be mistaken for Rett syndrome. It is important to rule out these conditions before diagnosing Rett syndrome.



Rett Syndrome



Management of Rett syndrome:

There is no known cure for Rett syndrome. A team approach requires open lines of communication between parents, physicians, therapists, educators and health and social agencies for the management of Rett syndrome.

Children with Rett syndrome demonstrate a far better ability to understand than they can express. They show a great desire to communicate through eye gaze, gestures and body language. A communication specialist can assist in developing an appropriate communication system. Music therapy has been shown to increase interest and responsiveness.

An occupational therapist will promote hand use with hand and elbow splints. They can design systems which allow the child to use assistive technology to access their environment.

Therapy is directed at maintaining and developing skills. Physical therapy encourages development of balance and walking. It minimizes deformities. This may include the use of braces, adaptive equipment, water therapy and therapeutic horse riding.

Parent Support:

The **Northwest Rett Syndrome Foundation** is a nonprofit organization founded by parents and professionals who recognized the need for a strong regional group.

Goals of the foundation:

- Facilitating early recognition of Rett syndrome.
- Promoting Rett syndrome awareness.
- Supporting research efforts in the Northwest.
- Hosting an annual conference for families and professionals.
- Creating parent-to-parent network.
- Providing updated information on current research and therapies through newsletters.
- Providing information on area resources and services.

You can help!

To learn more:

Northwest Rett Syndrome Foundation

PO Box 5475
 Salem, Oregon 97304
 1-888-326-2185
 Fax: 253-573-1966
 www.nwrettsyndrome.org
 Tax Exempt #93-0979668

Call For A Family Near You

1-888-326-2185



NWRSF has families from Alaska, Idaho, Northern California, Oregon, Washington & Western Canada

NW Rett Syndrome Foundation
 Dr. Sarojini Budden, Medical Advisor
 Dr. Patrick MacLeod, Medical Advisor
 Dr. Mario Petersen, Medical Advisor



Board Members 2009 (a few not pictured)

Understanding RETT SYNDROME



Conference Group Photo August 2008



N.W. Rett Syndrome Foundation