

## Goals & Purposes

### Rett Syndrome Association of Illinois

- To serve as a support system for families and professionals
- To promote an awareness and understanding of Rett syndrome
- To promote research of Rett syndrome
- To promote the general welfare of those whose lives are affected by Rett syndrome
- To host conferences, workshops and support group meetings
- To publish a newsletter
- To collect and disseminate information about Rett syndrome

To learn more about Rett syndrome and the Rett Syndrome Association of Illinois and to sign up for the *Rett Link* newsletter, visit our web site at [www.rettillinois.org](http://www.rettillinois.org).

### Rett Syndrome Association of Illinois

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Love is Caring & Sharing  
Founded 1989

# A Look at Rett Syndrome



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**Rett Syndrome  
Association of Illinois  
[www.rettillinois.org](http://www.rettillinois.org)  
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# Rett Syndrome

Rett syndrome is a neurodevelopmental disorder that begins in early infancy and affects primarily females, and very rarely males. The diagnosis of Rett syndrome is based on symptoms and clinical observation. The discovery of the *MECP2* gene has made possible the development of a blood test for Rett syndrome, which may be used to confirm a clinical diagnosis. The severity of symptoms may vary widely from patient to patient.

## Necessary for the Diagnosis of Rett Syndrome

- Period of apparent normal or near normal development until 6-18 months.
- Normal head size at birth followed by decreased rate of head growth from six months to four years in most children.
- Loss of purposeful hand use, replaced by stereotypical hand movements—may include hand washing/wringing/squeezing; hand clapping/tapping/rubbing; hand mouthing.
- Social withdrawal in some girls; communication dysfunction, loss of spoken words.
- Usually wide-based and stiff-legged gait or failing mobility.
- Impaired expressive/spoken language. Receptive language and cognitive abilities are difficult to assess.



# Diagnostic Criteria

## Supportive Criteria: May be seen in children with Rett syndrome (not needed for diagnosis)

- Breathing pattern irregularities which include hyperventilation, breath holding, apnea, air swallowing
- EEG abnormalities
- Seizures
- Scoliosis/kyphosis
- Teeth grinding
- Gastrointestinal issues which may include reflux, constipation, or gallbladder problems
- Growth retardation and decreased body fat and muscle mass
- Biting/chewing/swallowing difficulties
- Poor circulation to legs and feet; cold, purplish feet, usually growth impaired
- Shakiness of torso and/or limbs
- Decreased mobility with age
- Muscle rigidity/spasticity/joint contractures
- Abnormal sleep patterns
- Irritability and agitation
- Positive results for *MECP2* test



# Stages of Rett Syndrome

## **Stage I 6-18 months** Early Onset/Stagnation

This stage is usually overlooked, as the symptoms of Rett syndrome are just emerging and are somewhat vague. Infants may show less eye contact, disinterest in play activity; possible delays in gross motor milestones; deceleration of head growth; hypotonia.

## **Stage II 1-3 years** Rapid Destruction

Rapid regression; loss of previously acquired motor and verbal skills — including loss of purposeful hand use; irritability; sleep disturbances; social withdrawal and stereotypic hand movements.



## **Stage III 2 years +** Pseudo-stationary

Teeth-grinding; hyperventilation; gastrointestinal issues; difficulty assessing receptive language/cognitive function; scoliosis; increase in eye contact and communication.

## **Stage IV 10 years +** Late Motor Deterioration

Reduced mobility; intense eye gaze.