

# Rett Syndrome

## TIPS AND RESOURCES FOR FAMILIES



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### What is Rett syndrome?

Rett syndrome is a neurodevelopmental disorder that affects 1 in 10,000 females. Males with Rett syndrome typically do not survive infancy. The syndrome occurs across all racial, ethnic, and social groups. While development may begin at a typical pace, challenges related to communication, learning, and coordination usually will begin to appear within 6-18 months. Rett syndrome is often misdiagnosed as autism or cerebral palsy.

### What causes Rett syndrome?

Rett syndrome is caused by a mutation in the MECP2 gene on the X chromosome. MECP2 contains instructions for the production of a protein responsible for brain development. When insufficient amounts of this protein are produced, a number of other genes involved in development also will be affected.

### What are the effects of Rett syndrome?

Rett syndrome is generally described as having four stages. As stated above, infants often begin developing at a typical pace and then in the first stage (6-18 months) begin losing some skills. Infants and toddlers may lose the ability to make eye contact, sitting or crawling may be delayed, and growth may be slowed.

In Stage II (1-4 years of age), children often lose the ability to control their hands. Repeated hand wringing, hand washing, and clapping or tapping motions are common characteristics. Breathing challenges such as apnea and hyperventilation may occur. Motor movements may prove difficult and slowed head growth is often noticeable at this stage. A loss of interest in social interaction and communication may occur.

In stage III (2-10 years of age), the ability to perform motor functions may be lost. Seizures are common. However, many report that communication and alertness are improved during this stage.

Stage IV often includes muscle weakness and curvature of the spine. The ability to walk may be lost. However, repetitive hand movements and eye gaze tends to improve during this stage.

### Can Rett syndrome be treated?

While there is no cure for Rett syndrome, management of symptoms can greatly improve quality of life. Medications may be appropriate for breathing irregularities and to control seizures. Occupational therapy will assist children with skills like dressing and feeding. Physical therapy may improve and prolong motor movement. Speech-language therapy may help to improve both understanding and expression of speech verbally or through an assistive technology device.

### How will having a child with Rett syndrome affect my family?

It may be beneficial for a family with a member with Rett syndrome to seek group support. Membership in the International Rett Syndrome Foundation helps families get information on the latest treatments and therapies, and also helps them to connect with others in similar situations. Many issues experienced by people with Rett syndrome are similar to what people with autism spectrum disorders face, so membership in a local chapter of The Arc or Autism Society can be helpful for learning about special education law, local programs, and new therapies.

*Please see reverse for resources.*



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## RESOURCES

### Who We Are and Who We Serve

The **Vanderbilt Kennedy Center (VKC)** works with and for people with disabilities and their family members, service providers and advocates, researchers and policy makers. It is among only a few centers nationwide to be a University Center for Excellence in Developmental Disabilities, a *Eunice Kennedy Shriver* Intellectual and Developmental Disabilities Research Center, and a Leadership Education in Neurodevelopmental and Related Disabilities Training Program. The following are some of the ways the Center's programs and staff can assist families, educators, and other service providers.

### Tennessee Disability Pathfinder

Tennessee Disability Pathfinder is a free statewide phone, web, and print referral service in English and Spanish. It connects the Tennessee disability community with service providers and resources. Its website database has over 3,000 agencies searchable by Tennessee county and service. Pathfinder is a project of the VKC, TN Council on Developmental Disabilities, TN Department of Health, and the TN Department of Intellectual and Developmental Disabilities. Contact [www.familypathfinder.org](http://www.familypathfinder.org), (615) 322-8529, toll-free (800) 640-4636.

### Two Easy Ways to Take Part in Research

The Vanderbilt Kennedy Center serves families through research studies. StudyFinder is a searchable database that lists current VKC studies, including ASD research. Studies seek people of all ages with and without developmental disabilities. See [kc.vanderbilt.edu/studyfinder](http://kc.vanderbilt.edu/studyfinder), (615) 936-0448. Research Match is a secure place for volunteers and researchers to connect. Once you sign up and get added to the registry, a researcher will contact you if you're a possible match for the research study. See [www.researchmatch.org](http://www.researchmatch.org).

### Other Resources

- The Arc Tennessee  
Advocates for the rights and full participation of all people with intellectual and/or developmental disabilities.  
(800) 835-7077, [www.thearc.org](http://www.thearc.org)
- Family Voices of Tennessee  
A health information resource for families who have children with special health care needs: disability, chronic illness, physical or mental health conditions.  
(888) 643-7811, [www.tndisability.org/familyvoices](http://www.tndisability.org/familyvoices)
- Autism Society of East Tennessee  
(865) 247-5082, [www.asaetc.org](http://www.asaetc.org)
- Autism Tennessee  
(615) 385-2077, (866) 508-4987, [www.autismtn.org](http://www.autismtn.org)
- Autism Society of the Mid-South  
(901) 542-2767, [www.tnautism.org/west](http://www.tnautism.org/west)
- International Rett Syndrome Foundation  
(800) 818-7388, [www.rettsyndrome.org](http://www.rettsyndrome.org)
- Child Neurology Foundation  
(877) 263-5430, [www.childneurologyfoundation.org](http://www.childneurologyfoundation.org)
- Genetic and Rare Diseases Information Center  
(888) 205-2311  
<http://rarediseases.info.nih.gov/GARD/>

**Contact the Vanderbilt Kennedy Center**  
**Nashville (615) 322-8240**  
**Toll-Free (866) 936-VUKC [8852]**  
**Web: [vkc.vumc.org](http://vkc.vumc.org)**  
**Email: [kc@vumc.org](mailto:kc@vumc.org)**

