

Rett Syndrome

TIPS AND RESOURCES FOR FAMILIES



What is Rett syndrome?

Rett syndrome is a progressive neurodevelopmental disorder that is defined by:

- apparently typical development in the first 6 months of life,
- a period of developmental regression followed by stabilization,
- loss of spoken language,
- problems with walking,
- loss of purposeful hand use,
- and distinctive repetitive, hand movements (hand stereotypies).

Rett syndrome primarily affects girls and women and occurs in about 1 in 10,000 live female births. There have been rare reports of boys affected with symptoms consistent with Rett syndrome. The syndrome occurs across all racial, ethnic, and social groups. Early in its course, prior to development of the characteristic hand movements, Rett syndrome is often misdiagnosed as autism or cerebral palsy.

What causes Rett syndrome?

Rett syndrome is usually caused by a loss-of-function mutation in the X-linked gene *MECP2*. This gene is required for normal maturation of neurons and the

nervous system, and loss of the protein prevents the brain from maturing appropriately leading to widespread abnormalities in nervous system function. Only rarely is a genetic cause not identified in Rett syndrome.

What is the usual presentation of Rett syndrome?

After an uneventful pregnancy and delivery, most people with Rett syndrome have apparently typical development for the first 6 months of life. Between 6-18 months parents and/or physicians may note developmental delays that may be mild at first. Regression of spoken language and hand use with development of repetitive, purposeless hand movements are often the first distinctive signs of the syndrome usually occurring between 18 and 30 months of life. Loss of walking ability may also occur during this time. During the period of developmental regression, some individuals may have social withdrawal and/or irritability.

After the period of regression, a “plateau” stage occurs in which skills are no longer lost and may show slight improvement. This is also the period when other distinctive features may appear including irregular breathing patterns (hyperventilation, breath holding), poor sleep, problems with walking, cold hands and feet, and seizures. Problems coordinating chewing and swallowing, and additional gastrointestinal issues such as reflux and constipation are also very common. Although some individuals show “autistic” features during regression, during the plateau stage most show improved social interaction and eye contact.

During adolescence and young adulthood, movement may become more impaired, with slowness of movements, increased difficulty walking, and muscle stiffness. These clinical features are termed parkinsonism. Scoliosis may also develop during late childhood or early adolescence and may worsen with age.

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RESOURCES

Can Rett syndrome be treated?

The current approach to treatment is based on management of symptoms, which can greatly improve quality of life. This often requires input from multiple specialties including neurology, gastroenterology, rehabilitation medicine, orthopedics, and nutrition, with overall care being coordinated by a primary care provider. Medications may be used to control seizures, constipation, reflux, sleep problems, and movement disorders. Speech and occupational therapy will assist children with skills like chewing and swallowing, as well as maximize potential hand use. Physical therapy may improve and prolong walking and maximize range of motion. Despite limited verbal language, communication therapy may help to improve understanding and enhance communication through nonverbal methods including assistive technology devices such as computer-based eye-tracking systems. While there is currently no cure for Rett syndrome, the future does offer hope with multiple ongoing and planned clinical trials testing treatments with the potential to directly impact the course of the syndrome.

Vanderbilt Resources

- The **Vanderbilt Kennedy Center (VKC)** works with and for people with disabilities and their family members, educators and service providers, researchers, students, and policy makers. Faculty and staff engage in interdisciplinary research, training, service, and information dissemination and work in collaboration with local, state and national networks and partners. (615) 322-8240, toll-free (866) 936-8852, vkc.vumc.org
- The **Vanderbilt Rett Syndrome Program** is one of seven Rett Syndrome Research Trust Clinical Trial Consortium sites and is designated as a Rett Syndrome Clinical Research Center of Excellence. The clinical team has expertise in the evaluation, diagnosis, and treatment of children with Rett syndrome and related disorders. The research team leads innovative studies in mechanisms underlying Rett syndrome and related disorders, and aims to develop biomedical, pharmacological, and educational interventions to improve lives. Web: vkc.vumc.org/rett/

- **Tennessee Disability Pathfinder** provides free information, referral sources, and help with navigating services via phone, email, and website. Assistance is available to individuals of all ages, all types of disabilities, and all languages spoken. Its website includes a directory of more than 3,000 agencies searchable by Tennessee county, topic of interest, and other filters. Pathfinder is a project of the VKC and is partially funded by Tennessee Council on Developmental Disabilities and other state agencies. (615) 322-8529, toll-free (800) 640-4636, TNPathfinder.org

National Resources

Rettsyndrome.org is a national organization that provides the latest in medical information, funds research, offers meaningful resources and support, raises awareness, and engages in advocacy for individuals with Rett syndrome.

Web: rettsyndrome.org

Rett Syndrome Research Trust was launched in 2008 to drive research toward a cure for Rett syndrome. Families can register to stay informed on the latest Rett syndrome-related research and news.

Web: reverserett.org

Tennessee Resources

- **Family Voices of Tennessee**
tndisability.org/familyvoices
- **The Arc Tennessee**
theartn.org
- **Tennessee Technology Access Program (TTAP)**
tn.gov/humanservices/ds/ttap.html
- **Transition Tennessee**
transitiontn.org

