

## A Guide to Rett Syndrome

### ABOUT RETT SYNDROME

Rett syndrome (RTT) is a severe neurologic disorder that affects individuals in childhood. Occurring in approximately 1 in 10,000 female births and more rarely in males, it is caused by a mutation on the *MECP2* gene located on the X chromosome. Females have two X chromosomes; therefore, not all their cells are affected by the mutation. This cellular “mosaicism” adds to the complexity of Rett syndrome research. Additionally, over two hundred differing variants in the *MECP2* gene have been associated with Rett syndrome. <sup>(1)</sup>



RTT is characterized by a loss of acquired fine and gross motor skills and the development of severe neurologic and autonomic dysfunction. The hallmark of Rett syndrome is near-constant repetitive hand movements. Most individuals experience cardiac, respiratory, digestive, and muscular abnormalities. RTT can present with a wide range of disabilities ranging from mild to severe. They are often unable to walk or talk and many develop scoliosis and seizures. Cognitive assessment in children with Rett syndrome is complicated. Still, it is known that they understand far more than they can communicate to us, evidenced by their bright and attentive eyes and their ability to express a wide spectrum of moods and emotions. Those with RTT require lifelong care and can live well into adulthood.

In the United States, approximately 9,000 individuals are living with RTT. With the first treatment for Rett syndrome approved by the FDA on March 10, 2023, it is essential that our caregivers have access to this potentially life-changing medicine in their state of residence.

### INTERNATIONAL RETT SYNDROME FOUNDATION

*The mission of the nonprofit International Rett Syndrome Foundation (IRSF) is to accelerate full-spectrum research to cure Rett syndrome and empower families with information, knowledge, and connectivity.*

The International Rett Syndrome Foundation is a 501(c)3 organization founded in 2007. IRSF coordinates, cultivates, accelerates, and funds research that has the promise of being disease-modifying; ensuring that the lives of people living with this disorder are as meaningful as possible and free of discomfort. IRSF also supports families of individuals living with RTT and advocates to raise awareness about RTT. Learn more about IRSF’s mission at [rettsyndrome.org](https://rettsyndrome.org).

## CENTER OF EXCELLENCE NETWORK

International Rett Syndrome Foundation's Center of Excellence (CoE) Network consists of 18 Rett clinics nationwide that provide best-in-class clinical care. Clinics in the network meet a number of requirements that ensure that they can provide quality care to those with RTT. Each CoE has a physician director with expertise in RTT and health care services that meet the unique needs of individuals with RTT. These clinics remain involved in clinical trials and contribute to the IRSF Rett Syndrome Registry to increase understanding of RTT.



Typical CoE visits can include specialists in neurology, gastroenterology, genetics, physical medicine, and speech therapy. The CoE network is essential in providing cohesive care for individuals with RTT and ensuring that families and caregivers receive the tools they need to care for their loved ones.

## RESEARCH

With nearly 40 active research projects worldwide, the RTT community is closer than ever to specialized treatments and a cure. RTT is a complex disorder that requires multiple approaches. Because of this, IRSF invests in innovative research for new drugs to treat RTT and genetic treatments for curative approaches. The work doesn't end there. IRSF is also working to build a robust treatment pipeline and ensure clinical trial readiness and success. IRSF's strategy is simple but powerful: improve care today and create treatments for tomorrow. IRSF is committed to a full-spectrum approach to provide solutions for everyone living with Rett syndrome.

Rett syndrome research often contributes to the understanding, and possible treatments of other synaptic brain disorders, such as Alzheimer's, Autism, Fragile X, Parkinson's, and traumatic brain injury.

### Quote from a Rett Parent

*"We support IRSF because they have been a helpful resource for us. From Molly's diagnosis on, we have learned so much from the website and webinars! Rett is a scary diagnosis, but IRSF has helped us to feel empowered to provide the best support and care for Molly. We are also highly encouraged by the progress that doctors and scientists are making for Rett patients all over the world. It feels like a hopeful time to work hard to provide funding for our girls."* - **Kelsey Lahaie, mom to Molly**

## CONCLUSION

Although there is no cure for RTT, the International Rett Syndrome Foundation has an established process that allows individuals with RTT to live meaningfully with the support of clinicians who have dedicated procedures to care for this complex disorder. Researchers are exploring all possible avenues to treat and cure RTT, and they remind our community that there is hope for their loved ones.

