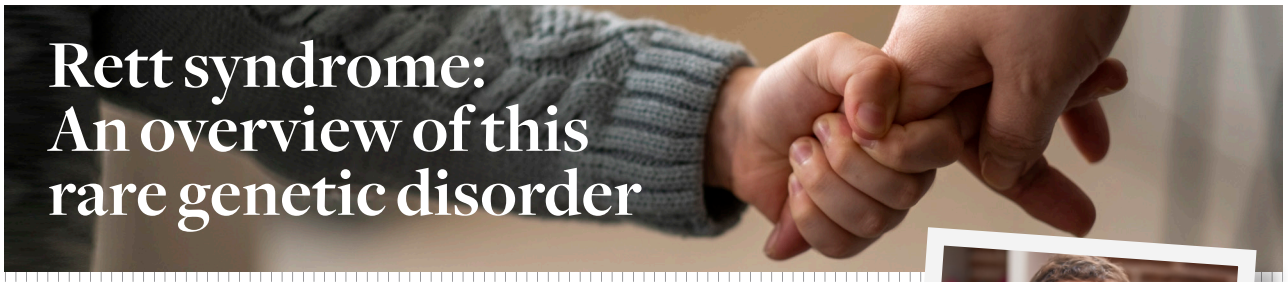
Rett syndrome: An overview of this rare genetic disorder



What is Rett syndrome?

Rett syndrome is a rare genetic disorder in which a child's early growth and development regresses after initially meeting their developmental milestones. It is a neurodevelopmental disorder, which means it affects how the brain and nervous system develop. As a result, the child can have severe communication or coordination problems. Children with Rett syndrome may behave or move similarly to children with autism spectrum disorder, which is another neurodevelopmental disorder.

Rett syndrome can begin as early as 6 months of age, with symptoms being so subtle that parents may not even notice the change in their child's development.

Symptoms

Symptoms of Rett syndrome usually do not appear right away. Children with the disorder may meet age-appropriate milestones for the first 6 to 18 months before they start showing symptoms. Some common symptoms are:

- Loss of speech
- Loss of hand movements such as grasping
- Compulsive movements such as hand wringing
- Balance problems
- Breathing problems
- Social or behavior problems
- Learning problems or intellectual disability

Who is more likely to have Rett syndrome?

Although Rett syndrome is a genetic disorder, most cases occur randomly—the chance that someone will inherit Rett syndrome is less than 1%. Children from all racial and ethnic groups may develop Rett syndrome, with most cases diagnosed in girls. It is caused by a mutation in the MECP2 *(methyl CpG binding protein 2)* gene, which is located on the X chromosome and contributes to brain development. Some girls with Rett syndrome also have female family members with the MECP2 gene mutation.

Not everyone with a MECP2 gene mutation will develop Rett syndrome.

How can doctors find out whether a child has Rett syndrome?

There are prenatal screenings available to find a MECP2 gene mutation, but the same gene mutation in Rett syndrome can be found in other conditions. It is important to notify a doctor if a child shows behavioral changes or physical problems, especially if these changes happened after previously age-appropriate development. That can help a pediatric neurologist, clinical geneticist, or developmental pediatrician screen for and diagnose the disorder.

Is there a cure?

Not at this time. Treatments are being developed to help patients with Rett syndrome manage their symptoms. These may include medications, occupational therapy, special equipment to help children with physical movement, nutritional programs, and educational and social support. At NIH, the National Institute of Neurological Disorders and Stroke, the *Eunice Kennedy Shriver* National Institute of Child Health and Human Development, the National Institute of Mental Health, and the Division of Rare Diseases Research Innovation at the National Center for Advancing Translational Sciences all support research on Rett syndrome.

The Rett Syndrome Research Trust supports clinical research into treatments and cures for the disorder. The Trust also maintains a digital Rett Syndrome Global Registry for parents and caregivers to share data with doctors. The goal is to help track treatment results and research discoveries around Rett syndrome. ■

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