



Rett syndrome (RTT), Causes and Treatment

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Rett syndrome (RTT) is a rare genetic condition that results in certain mental and physical disabilities. It tends to only affect females.

RTT develops due to a mutation in one of the genes on the X chromosome. The gene in question is the methyl CpG binding protein 2 (MECP2) gene.

RTT is very rare and occurs in around 1 in every 10,000–15,000 live female births. There are four stages of RTT, and the first stage typically begins between the ages of 6 and 18 months.

RTT is a neurological condition. An infant with RTT will usually have normal early growth at the beginning of life, followed by a number of symptoms that develop in stages.

Infants with RTT may have one or more of these common symptoms:

- slowing of development
- loss of purposeful use of the hands
- distinctive hand movements
- slowed brain growth
- slowed head growth
- difficulty walking
- seizures
- intellectual disability

The condition primarily affects females. This is because the MECP2 gene is on the X chromosome. Females have two X chromosomes, while males only have one X chromosome. In females, one of each two X chromosomes is inactive in each cell.

Which one is active is random, which is why the severity of RTT varies so much from person to person.

In males, there is only one X chromosome, so RTT is typically lethal to the male fetus. If the fetus does survive to birth, the infant will likely have a severe case of RTT and likely die early in life.

Treatments

There is currently no cure for RTT, and the reason behind the mutations that cause it is still largely unknown.

For this reason, treatment options for RTT are limited. Most treatment focuses on symptom relief.

Treatment options that can help a person with RTT include:

- **Medications:** Medications can help treat breathing irregularities and motor difficulties. A doctor may also prescribe drugs to help control seizures.
- **Physical therapy and hydrotherapy:** This can help a person with their mobility issues and balance.
- **Occupational therapy:** Occupational therapy can help a person develop the skills they need to perform tasks such as dressing and feeding.
- **Speech and language therapy:** This can help a person learn how to use nonverbal communication and improve their social interaction.
- **Braces:** These may be helpful for correcting the effects of scoliosis.
- **Splints:** A doctor may use splints to help adjust hand movements.
- **Nutritional help:** Supplements can strengthen the bones and assist with scoliosis. Having a specific nutritional program may help the person maintain a moderate weight.