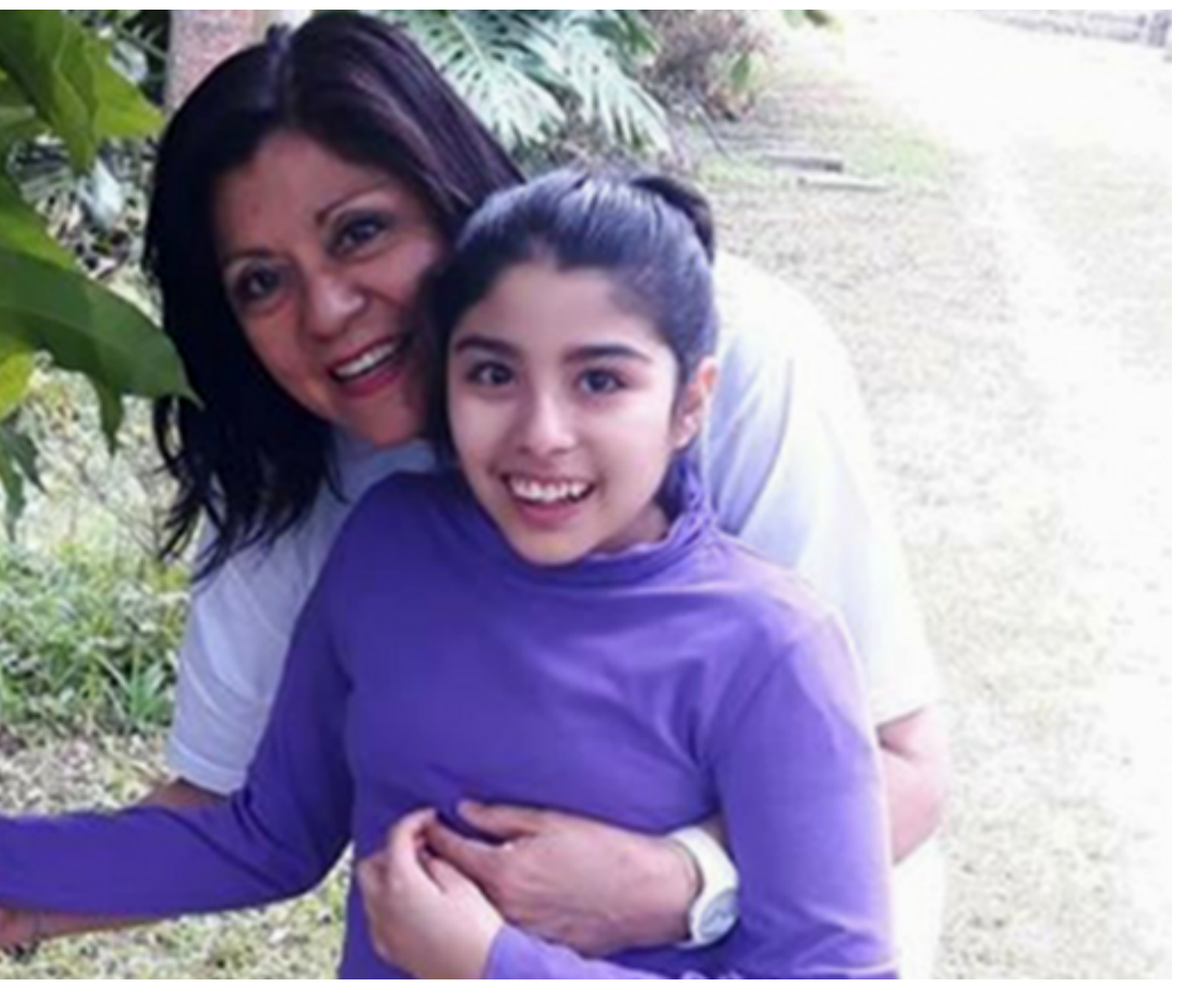
Fact Sheet :

Rett Syndrome

apd
WESTERN CAPE ASSOCIATION FOR PERSONS WITH DISABILITIES
TM: WCAPD



What is Rett Syndrome?

Rett syndrome is a rare genetic neurological disorder that occurs almost exclusively in girls and leads to severe impairments, affecting nearly every aspect of the child's life: their ability to speak, walk, eat, and even breathe easily. The hallmark of Rett syndrome is almost constant repetitive hand movements.

Children with Rett syndrome often have normal development during the first 6 to 18 months of life. After this time, they have a period of regression (loss of skills) and they may lose speech and other developmental milestones. The symptoms can range from mild to severe.

Rett syndrome is caused by mutations on the X chromosome on a gene called MECP2. It occurs worldwide in 1 of every 10,000 female births, and is even rarer in boys.

Rett & Angelman syndromes have similar clinical, neurological, and behavioral characteristics, but they do not appear to share similar facial features.

Signs & symptoms

While many syndromes are defined by characteristic facial features, Rett syndrome is defined traditionally by changing behaviour and developmental composition, which occurs after an apparently normal early infant period.

- Loss of speech
- Loss of purposeful use of hands
- Involuntary hand movements such as handwashing
- Loss of mobility or gait disturbances
- Loss of muscle tone
- Seizures or Rett "episodes"
- Scoliosis
- Breathing issues
- Sleep disturbances
- Slowed rate of growth for head, feet and hands

Quick questions

Is there a cure?

No there is no cure, but research is underway on new drugs that may improve management of symptoms..

Can children go to school?

Your child should be able to attend school, but may need special accommodations. Getting early treatment, including physical therapy, speech therapy, and occupational therapy, can help your child learn and communicate.

What is the life expectancy?

Due to the rarity of RTT, very little has been published about life expectancy. Data from the Natural History Study have showed that a girl with RTT has a 100% chance of reaching age 10, a 90% chance of reaching age 20, a greater than 75% chance of reaching age 30, a greater than 65% chance of reaching age 40, and a greater than 50% chance of reaching age 50. As improved nutrition and overall care are provided, these probabilities are expected to improve.

Western Cape Association for Persons with Disabilities
www.wcapd.org.za
0215552881

Resources : Boston Children's Hospital
RettSyndrome.org
Mayo Clinic