

Symptoms and Causes of Rett Syndrome

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DESCRIPTION

Rett syndrome is a neurological and developmental illness caused by a rare genetic mutation that changes the way the brain develops. This condition causes movement and linguistic skills to deteriorate over time. Females are the ones that are most affected by Rett syndrome. For the first six months of life, most baby girl with Rett syndrome appear to develop normally. These babies then lose the capacity to crawl, walk, talk, and use their hands, among other things. Children with Rett syndrome develop issues using muscles that control movement, coordination, and communication throughout time. Seizures and intellectual difficulties are also possible side effects of Rett syndrome.

Unusual hand gestures, such as rubbing or clapping repeatedly, have taken the place of deliberate hand use. Although there is no cure for Rett syndrome, researchers are looking into possible therapies. Treatment for children and people with Rett syndrome and their families currently focuses on increasing movement and speech, controlling seizures, and providing care and support.

Rett syndrome is caused by mutations on the X chromosome on a gene called *MECP2*. *MECP2* gene, there are over 900 different mutations. The majority of these mutations are concentrated in eight distinct "hot areas." Rett syndrome is not a degenerative disease, and most people who have it live to be middle-aged or older. Rett syndrome affects one out of every 10,000 female babies born around the world, and it's extremely rarer in boys. Rett syndrome can cause a variety of symptoms, ranging from minor to severe. The location, kind, and severity of the mutation, as well as X-inactivation, affect the course and severity of Rett syndrome.

The age at which Rett syndrome shows itself, as well as the severity of symptoms, differs from kid to child. However, before the symptoms occur, the child appears to grow and develop normally, though there are typically minor abnormalities, such as loss of muscle tone (hypotonia), difficulties eating, and jerkiness in limb motions, even in early infancy. Then, over time, mental and physical symptoms begin to emerge. The child's ability to communicate and meaningful use of her hands deteriorates as the syndrome grows.

Other early signs and symptoms include difficulty crawling or walking, as well as a loss of eye contact. Compulsive hand movements such as wringing and washing accompany the loss of functional use of the hands. The commencement of this period of regression can be rapid in some cases.

Symptoms

Rett syndrome symptoms, progression, and severity can differ considerably from one person to the next. The illness typically affects women and is thought to be part of a larger group of diseases linked to mutations in the *MECP2* gene. Rett syndrome has the potential to cause a wide range of disabilities. Symptoms usually manifest in stages. It's crucial to note that people who are impacted may or may not experience all of the symptoms listed below. Affected persons should discuss their specific condition, related symptoms, and overall prognosis with their physician and medical team. The age at which symptoms arise vary, most babies with Rett syndrome appear to grow normally for the first six months before showing any evidence of the illness.

The most typical alterations occur between the ages of 12 and 18 months, and they might occur suddenly or gradually. The brain does not develop normally, and the head is typically tiny (doctors call this microcephaly). As the youngster gets older, the child's stunted growth becomes more obvious. Most of the symptoms seen as:

- Hand movements are causing problems. The majority of youngsters with Rett syndrome lose their ability to use their hands. They have a habit of rubbing or wringing their hands together
- There are no linguistic skills. Between the ages of one and four, social and language abilities begin to deteriorate. Rett syndrome causes children to cease talking and have severe social anxiety. They may avoid or be uninterested in other people, toys, and their environment
- Muscle twitching and coordination issues. Walking becomes difficult as a result of this
- Speech impairment
- Hands are no longer used for their intended purpose
- Hand movements that are not voluntary, such as hand-washing
- Gait abnormalities or loss of mobility

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- Muscle tone declines
- Rett "episodes" or seizures
- Scoliosis
- Breathing problems
- Disruptions in sleep
- Growth of the head, feet, and hands has slowed