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Opinion Article

Rett Syndrome: Stages and how it Impact on People

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DESCRIPTION

Rett Syndrome is an uncommon, serious neurological condition that primarily affects females. Rett syndrome is typically diagnosed in a child's first two years of life, and it can be a frightening diagnosis for parents. Rett syndrome has no known treatment or cure, however early detection and intervention may benefit girls and their families. It was once thought to be a component of autism spectrum disorder. We now understand that it is primarily genetic in nature.

A child with Rett syndrome may experience uncoordinated breathing and seizures, including hyperventilation—rapid, forceful inhalation and exhalation of air or saliva—and air swallowing. As age, kids with Rett syndrome also have a propensity to grow tight and irritable. They might have protracted bursts of laughing or cry or scream for long stretches of time. Rett syndrome symptoms typically don't get better with time. It lasts the rest of one's life. The symptoms frequently don't change or instead slowly get worse. People with Rett syndrome rarely have the ability to live freely.

A mutation on the X chromosome is present in the majority of Rett syndrome sufferers. It is unclear exactly what this gene accomplishes or how a mutation causes Rett syndrome. Numerous genes involved in development may be influenced by the same gene, according to researchers. Rett syndrome is inherited, but kids hardly get the bad gene from their parents. Instead, a DNA mutation occurs accidentally. Rarely will boys with the Rett syndrome mutation survive past birth. Males only have one X chromosome, as opposed to the two that females do, hence the disease's effects are significantly more severe and almost usually deadly.

Stages of the disorder

Rett syndrome normally has four stages.

Stage I: It also known as early onset, often starts between the ages of 6 and 18 months. This period is frequently missed since the disorder's symptoms can be hazy, and parents and medical professionals might not first detect the child's growth slowing down. The baby can start to make fewer eye contacts and show less interest in toys. Gross motor skills like sitting or crawling could be delayed. There may be hand-wringing and a slowdown in head growth, but not enough to get notice. Though it seldom lasts longer than a year, this stage often lasts a few months.

Stage II: It often known as the quick destructive stage, typically starts between the ages of 1 and 4 and can endure for several weeks or even months. As the youngster loses intentional hand skills and spoken language, it may start abruptly or gradually. This period is frequently when distinctive hand gestures like wringing, washing, clapping, or tapping, as well as repeatedly moving the hands to the lips, start. The child's hands may be clasped behind his or her back or held at the sides while being randomly touched, grabbed, and released. While the child is awake, the movements continue, but stop when they are asleep. Although breathing often gets better as you sleep, breathing irregularities such apneic episodes and hyperventilation may still happen. Additionally, some girls exhibit traits like autism, such as a lack of social skills and communication. Initiating motor actions and walking may be challenging. During this period, the growth of the head is typically seen to slow.

Stage III: The plateau or pseudo-stationary stage, often starts between the ages of 2 and 10 and can remain for years. During this stage, apraxia, motor issues, and seizures are common. However, there can be a change in the child's behavior, with reduced weeping, impatience, and autistic-like traits. A female in stage III might become more aware, pay attention for longer periods of time, and communicate more effectively. For the majority of their life, many females stay in this stage.

Stage IV: It often known as the late stage of motor degeneration, can endure for years or even decades. Reduced mobility, scoliosis, and muscle weakness, rigidity, spasticity, and increased muscle tone are notable characteristics, as is abnormal posture of an arm, leg, or top section of the body. Girls who could previously walk might stop doing so. In stage IV, cognitive, communicative, or hand abilities normally do not deteriorate. Eye focus normally improves while repetitive hand movements may be reduced.

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