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Short Communication

Rett Syndrome Complications and their Symptoms

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DESCRIPTION

Rett Syndrome (RTT) is a hereditary condition that commonly manifests in females between the ages of 6 and 18 months. The symptoms include repetitive movements, linguistic problems, and coordination issues. Those who are affected frequently grow more slowly, have trouble walking, and have smaller heads. Seizures, scoliosis, and sleep issues are some of the Rett syndrome's complications [1]. There are differences in the condition's severity.

A genetic mutation in the X chromosome's *MECP2* gene causes Rett syndrome. Less than 1% of cases are inherited from a person's parents, making it virtually always a novel mutation [2]. It nearly only affects females; males with a comparable mutation often pass away soon after birth. Genetic testing can be used to confirm the diagnosis, which is based on the symptoms. Rett syndrome has no known treatment. Anticonvulsants might be used to treat seizures. Braces, physiotherapy, and special education may all be beneficial. Many people with the illness live into their middle years.

Signs and symptoms

Stage I: Stage I, often known as early-onset, typically starts between the ages of 6 and 18 months. Because the disorder's symptoms may be hazy, parents and medical professionals may first fail to notice the child's growth slowing down [3]. 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: Stage II, 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 stage, which is referred to as mouthing, is frequently when distinctive hand motions like wringing, washing, clapping, or tapping, as well as repeatedly moving the hands to the mouth, 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 [4]. The child's movements continue when they are awake but stop while they are asleep. Although breathing often gets better as you sleep, breathing irregularities such apneic episodes and hyperventilation can still happen. Some girls also exhibit traits like autism, including a lack of social contact and speech. Walking may be shaky, and starting motor actions may be challenging. During this period, the growth of the head is typically seen to slow.

Stage III: The plateau or pseudo-stationary stage, also known as Stage III, 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. In stage III, there may be an increased interest in the environment as well as an increase in awareness, attention span, and communication abilities. For the majority of their life, many females stay in this stage [4].

Stage IV: Stage IV, often known as the late stage of motor degeneration, can endure for years or even decades. Notable characteristics include decreased mobility, spinal curvature, and muscle weakness, rigidity, spasticity, and increased muscle tone along with aberrant arm- or leg-posture. Girls who could previously walk might stop doing so. In stage IV, cognitive, communicative, or hand abilities typically do not deteriorate. Eye focus normally improves, and repetitive hand movements may be reduced.

CONCLUSION

Rett Syndrome (RTT) is a hereditary condition that is brought on by random or germline mutations in the *MECP2* gene on the X chromosome, which is involved in transcriptional silence and epigenetic regulation of methylated DNA. Less than 10% of RTT cases have been found to also have similar mutations in the CDKL5 or FOXG1 genes. Rett syndrome is originally identified through clinical observation, but a genetic *MECP2* gene deficiency renders the diagnosis conclusive.

Rett syndrome has been characterized as a neurodevelopmental disorder rather than a neurodegenerative one. One indication of this is the lack of neuronal death observed in mice with

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generated Rett Syndrome. Additionally, some research suggests that adult mice with these traits may have some of their symptoms partially recovered by reintroducing functioning *MECP2* gene. This knowledge has also influenced subsequent research projects aimed at curing the condition.

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