OPEN ACCESS Freely available online

Autism: Open Access

Short Communication

Rett Syndrome: Its Clinical Presentations and Research Efforts in Several Cognitive and Motor Impairments

Komal Zade*

Department of Genetics, University of Trinity, Dublin, Ireland

DESCRIPTION

Rett Syndrome is a rare and complex neurodevelopmental disorder that primarily affects females. Named after Austrian pediatrician Andreas Rett, who first described the condition in 1966, Rett Syndrome is characterized by a distinctive pattern of regression in development, leading to severe cognitive and motor impairments. This article delves into the various aspects of Rett Syndrome, including its clinical presentation, genetic basis, diagnosis, treatment options, and the ongoing research efforts aimed at understanding and managing this challenging disorder.

Clinical presentation

Rett Syndrome typically manifests in early childhood, with initial development appearing normal during the first six to eighteen months of life. However, between the ages of six months to two years, a regression in development becomes apparent. Affected children start to lose acquired speech and motor skills, develop hand-wringing movements, and exhibit social withdrawal. This regression phase is often distressing for parents and caregivers, as they witness their child's abilities decline [1].

The hallmark feature of Rett Syndrome is the loss of purposeful hand skills, replaced by repetitive and stereotypical hand movements, such as hand-wringing, clapping, and tapping. Other common symptoms include breathing irregularities, seizures, scoliosis, and motor impairments. It's important to note that the severity and range of symptoms can vary widely among individuals with Rett Syndrome.

Genetic basis

Rett Syndrome is primarily caused by mutations in the Mobile Electronics Certified Professional (*MECP2*) gene, located on the X chromosome. Most cases of Rett Syndrome arise spontaneously due to *de novo* mutations, meaning they are not inherited from parents. This genetic mutation affects the regulation of other genes in the brain, leading to the characteristic symptoms of the disorder.

MECP2 mutations are not limited to a single type, and different mutations can result in varying degrees of severity. Some mutations are associated with classic Rett Syndrome, while others may lead to atypical forms of the disorder, with different clinical presentations and outcomes [2].

Diagnosis

Diagnosing Rett Syndrome can be challenging, as its symptoms often overlap with other neurodevelopmental disorders like autism and cerebral palsy. A clinical diagnosis is typically based on the presence of specific criteria outlined in the Diagnostic and Statistical Manual of Mental Disorders (DSM-5). Genetic testing, particularly for MECP2 mutations, is a crucial component of confirming the diagnosis [3].

Treatment and management

Currently, there is no cure for Rett Syndrome. However, various therapies and interventions can help manage the symptoms and improve the quality of life for individuals with the disorder. These include:

Physical and occupational therapy: These therapies focus on improving mobility, muscle strength, and functional skills, helping individuals with Rett Syndrome maintain as much independence as possible.

Speech therapy: While many individuals with Rett Syndrome lose the ability to speak, speech therapy can help them communicate using alternative methods, such as Augmentative and Alternative Communication (AAC) devices.

Medications: Medications may be prescribed to manage specific symptoms, such as seizures and breathing irregularities. However, these treatments are typically symptomatic and do not address the underlying cause.

Supportive care: Providing a supportive and nurturing environment is essential for individuals with Rett Syndrome. This includes specialized education, social services, and assistance with daily living activities.

Correspondence to: Komal Zade, Department of Genetics, University of Trinity, Dublin, Ireland, E-mail: komal_zade@tedu.com

Received: 29-Aug-2023, Manuscript No. AUO-23-26825; **Editor assigned:** 01-Sep-2023, PreQC No. AUO-23-26825 (PQ); **Reviewed:** 15-Sep-2023, QC No. AUO-23-26825; **Revised:** 22-Sep-2023, Manuscript No. AUO-23-26825 (R); **Published:** 29-Sep-2023, DOI: 10.35248/2165-7890.23.13.383

Citation: Zade K (2023) Rett Syndrome: Its Clinical Presentations and Research Efforts in Several Coginitive and Motor Impairments. Autism: Open Access. 13:383.

Ongoing research

Research into Rett Syndrome is ongoing, with a focus on understanding the underlying biology of the disorder and developing potential treatments. Several promising avenues of investigation include:

Gene therapy: Scientists are exploring gene therapy approaches to correct the MECP2 mutations responsible for Rett Syndrome, with the aim of restoring normal gene function.

Drug therapies: Clinical trials are underway to test the effectiveness of various drugs in alleviating specific symptoms of Rett Syndrome, such as breathing abnormalities and motor impairments.

Biomarker discovery: Identifying biomarkers associated with Rett Syndrome could aid in early diagnosis and tracking disease progression, potentially leading to more targeted treatments [4].

CONCLUSION

Rett Syndrome is a rare and complex neurodevelopmental disorder that profoundly impacts the lives of affected individuals and their families. While there is currently no cure for Rett Syndrome, ongoing research offers hope for improved treatments and a better understanding of the underlying mechanisms of the disorder. In the meantime, early diagnosis and access to appropriate therapies and support services remain essential in enhancing the quality of life for individuals living with Rett Syndrome. Increased awareness, continued research, and support for affected families are crucial in the quest for a brighter future for those touched by this challenging condition.

REFERENCES

1. Amankwaa LC, Pickler RH, Boonmee J. Maternal responsiveness in mothers of preterm infants. Int Breastfeed J. 2007;7(1):25-30.
2. Deconinck N, Soncarrieu M, Dan B. Toward better recognition of early predictors for autism spectrum disorders. Pediatr Neurol. 2013;49(4):225-231.
3. Fernald A, Kuhl P. Acoustic determinants of infant preference for motherese speech. Sci Rep. 1987;10(3):279-293.
4. Sherrod KB, Crawley S, Petersen G, Bennett P. Maternal language to prelinguistic infants: Semantic aspects. J Acoust Soc Am. 1978;1:335-345.