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Perspective

Angelman Syndrome (AS) Impact on Individuals by Cognitive and Behavioral Functioning

Ernest Blank*

Department of Genetics, University of Makerere, Kampala, Uganda

DESCRIPTION

Angelman Syndrome (AS) is a rare neurodevelopmental disorder that affects approximately 1 in 12,000 to 20,000 individuals globally. First described by Dr. Harry Angelman in 1965, this syndrome presents with a distinct set of characteristics that impact neurological development, behavior and cognitive abilities. Despite its rarity, Angelman syndrome has garnered significant attention from researchers and medical professionals due to its unique features and challenges for those affected and their families.

Characteristics of angelman syndrome

Individuals with Angelman syndrome often exhibit a distinct phenotype characterized by developmental delays, intellectual disabilities and neurological abnormalities. Some of the indicated characteristics of Angelman syndrome include:

Developmental delays: Children with Angelman syndrome typically experience delays in reaching developmental milestones such as sitting, crawling and walking. Speech development is also significantly affected and many individuals with AS may never develop functional speech.

Intellectual disabilities: Individuals with Angelman syndrome usually have moderate to severe intellectual disabilities. They may have difficulties with learning, problem-solving and adaptive functioning.

Movement and balance issues: Many individuals with AS have problems with movement and balance, often presenting with ataxic gait and tremors. These motor impairments can significantly impact their ability to perform daily activities independently.

Seizures: Seizures are a common feature of Angelman syndrome, with onset usually occurring in early childhood. These seizures may vary in severity and frequency among affected individuals.

Distinct behavioral characteristics: Individuals with Angelman syndrome often exhibit a unique behavioral profile characterized by frequent laughter, smiling and excitability. They may also display hyperactivity, short attention spans and sleep disturbances.

Causes of angelman syndrome

Angelman syndrome is primarily caused by genetic abnormalities involving the 15q11.2-q13 region of chromosome 15. The majority of cases (about 70-75%) result from the deletion of a segment of the maternal chromosome 15 in this region. In other cases, AS may be caused by mutations in the *Ubiquitin Protein Ligase E3A (UBE3A)* gene, which is located on chromosome 15 and is critical for normal brain function.

In rare instances, Angelman syndrome may result from uniparental disomy, where both copies of chromosome 15 are inherited from the father or from imprinting defects that disrupt the normal expression of genes in the 15q11.2-q13 region. These genetic abnormalities lead to the absence or dysfunction of the UBE3A protein, which plays a crucial role in synaptic function and neuronal communication in the brain.

Management and treatment

While there is currently no cure for Angelman syndrome, management strategies focus on addressing the specific symptoms and improving the quality of life for individuals with AS and their families. A multidisciplinary approach involving medical professionals, therapists, educators and caregivers is often essential in providing comprehensive care and support.

Early intervention: Early intervention programs that include speech therapy, physical therapy, occupational therapy and behavioral interventions can help address developmental delays and improve functional abilities in children with Angelman syndrome.

Seizure management: Since seizures are common in individuals with AS, appropriate seizure management strategies, including medication and close monitoring are essential in minimizing seizure frequency and severity.

Behavioral support: Behavioral interventions and support services can help manage the unique behavioral characteristics

Correspondence to: Ernest Blank, Department of Genetics, University of Makerere, Kampala, Uganda, Email: ernest_blank@usedu.com

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associated with Angelman syndrome. Strategies such as Applied Behavior Analysis (ABA) and Positive Behavior Support (PBS) may be beneficial in addressing challenging behaviors and promoting social skills development.

Educational support: Individuals with Angelman syndrome may benefit from Individualized Education Plans (IEPs) and specialized educational programs made to their specific needs and abilities. Educators and school personnel can work collaboratively with families to create supportive learning environments and facilitate academic progress.

Supportive care: Ongoing medical monitoring and management of coexisting conditions such as sleep disorders, gastrointestinal issues and orthopedic problems are important aspects of comprehensive care for individuals with Angelman Syndrome.

CONCLUSION

Angelman syndrome is a complex neurodevelopmental disorder characterized by a unique constellation of features that impact individuals' physical, cognitive and behavioral functioning. While there is currently no cure for AS, advances in research and clinical care have led to improved understanding and management of the condition. By implementing comprehensive support and intervention strategies, individuals with Angelman syndrome can achieve their fullest potential and lead fulfilling lives within supportive and nurturing environments. Continued research efforts aimed at resolving the underlying genetic mechanisms and developing targeted therapies hold assurance for improving outcomes and enhancing the quality of life for individuals and families affected by Angelman syndrome.