

Angelman Syndrome: Causes, Symptoms and Treatment

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DESCRIPTION

Angelman syndrome is a rare genetic disorder that affects the nervous system, causing severe developmental disabilities and neurological problems. This disorder is characterised by delayed development, intellectual incapacity, significant speech impairment, and balance and movement issues (ataxia). The majority of affected children also suffer epilepsy and microcephaly, which is small heads. By the age of 6 to 12 months, delayed development is apparent, and other common signs and symptoms typically start to show up in early infancy. Children with Angelman syndrome frequently smile, laugh, and flail their hands about in an upbeat, energetic manner. Short attention spans and hyperactivity are prevalent. The majority of affected youngsters also has trouble falling asleep and requires less sleep than usual.

Causes

Angelman syndrome is primarily caused by a genetic mutation or deletion in a specific region of chromosome 15. There are different genetic mechanisms that can lead to Angelman syndrome, including:

Deletion: Approximately 70% of individuals with Angelman syndrome have a deletion on the maternally inherited chromosome 15. This means that a part of the genetic material is missing.

Paternal uniparental disomy: About 2-5% of individuals with Angelman syndrome have both copies of chromosome 15 inherited from the father, instead of one from each parent. This results in the absence of the maternal copy of the *UBE3A* gene, which is responsible for the syndrome.

***UBE3A* gene mutation:** In some cases, individuals may have a mutation in the *UBE3A* gene, which is responsible for producing a protein involved in brain development and function. This mutation prevents the gene from functioning correctly, leading to Angelman syndrome.

Symptoms

The symptoms of Angelman syndrome can vary in severity among individuals, but they typically include:

Developmental delays: Delayed or absent speech, motor milestones (such as sitting, walking), and intellectual disabilities are common. Most individuals with Angelman syndrome have severe to profound intellectual disability.

Movement and balance issues: Individuals with Angelman syndrome often have difficulty with balance and coordination, resulting in an unsteady or jerky gait. They may also have hand flapping movements or exhibit frequent laughter or smiling.

Seizures: Epilepsy is prevalent among individuals with Angelman syndrome. Seizures may start in early childhood or infancy and can vary in type and frequency.

Communication and behavioral difficulties: Limited or absent speech is a characteristic feature of Angelman syndrome. Communication challenges are often supplemented with alternative methods such as gestures, signs or Augmentative and Alternative Communication (AAC) systems. Individuals may also have hyperactivity, attention deficit disorder, and a happy demeanor with frequent laughter.

Sleep disturbances: Many individuals with Angelman syndrome experience abnormal sleep patterns, including a decreased need for sleep, frequent nighttime awakenings, and difficulty falling asleep.

Treatment

There is currently no cure for Angelman syndrome, and treatment focuses on managing the symptoms and improving the individual's quality of life. This may include:

Physical therapy: These therapies can help improve motor skills, coordination and overall physical development.

Speech therapy: Speech therapy can assist with communication skills, including non-verbal forms of communication.

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Medications: Seizures sleep disturbances and behavioral issues can be managed with medications prescribed by a healthcare professional.

Behavioral interventions: Strategies such as positive reinforcement, structured routines, and specialized education programs can help address behavioral challenges and promote learning and development.

Supportive care: Supportive care from a multidisciplinary team, including medical specialists, therapists, and educators, is crucial for managing the unique needs of individuals with Angelman syndrome and supporting their families.