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Commentary

Diagnosis and Treatment Involved in Angelman's Syndrome

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DESCRIPTION

A hereditary condition is called Angelman's syndrome. It results in intellectual disability, speech and balance issues, developmental delays, and occasionally seizures. Angelman's syndrome patients usually have joyful, vivacious dispositions and regularly grin and laugh. It is very rare in born babies. The life expectancy is normal but the diseased patients need support throughout the life. Most of the people with Angelman's syndrome don't have a family history of the disease.

Signs and symptoms of Angelman's syndrome include: developmental delays, such as failure to babble or begin to crawl by 6 to 12 months, intellectual handicap, little or nonexistent speaking, difficulty moving, balance, or walking, frequently grinning and giggling, a cheerful, vivacious personality, feeding or sucking challenges, difficulty falling and staying asleep.

The following characteristics may be present in people with Angelman's syndrome such as Seizures, which often start between the ages of 2 and 3, jerky or stiff movements, small head size and back of the head flatness, With the tongue out, Light-colored eyes, complexion, and hair, Unusual actions, such as walking with arms raised and hands flapping, issues with sleep, bent spine (scoliosis).

The complications associated with Angelman's syndrome may include feeding difficulties, hyperactivity, sleep disorders, obesity occurs in older children and scoliosis.

Diagnosis

Some tests are performed by a physician to diagnose the Angelman's syndrome using the following techniques:

Parenteral DNA pattern: Three of the four known genetic disorders that cause Angelman's syndrome are screened for by this test, known as a DNA methylation test.

Absence of chromosomes: A chromosomal microarray (CMA) can demonstrate the portions of missing chromosomes, if any chromosomes are missing.

Gene mutation: Rarely, a person's maternal copy of the UBE3A gene may be active but mutated, resulting in Angelman'ssyndrome. A UBE3A gene sequencing test to check for a maternal mutation may be prescribed by physician, if results from a DNA methylation test are normal.

Medical history: Angelman's syndrome patients are born without any developmental problems. The medical history of children's can help their doctor to determine the other conditions.

Physical examination: The physician will look for distinct physical characteristics such a large head, crossed eyes, or deep-set eyes.

Blood test: This involves genomic assessments that look out genes which are absent or missing.

Treatment

The treatment is based on the symptoms of Angelman's syndrome. These include:

Anti-seizure medication: Anticonvulsants is known as anti-seizure drugs which are used to manage or control seizures.

Sedatives: These are useful for the treatment of sleep disorders.

Medicines for agitation: These medications facilitate the digestion of food.

Physical treatment: Ankle braces and physical therapy are helps to improve mobility.

Occupational therapy: A specialist might provide techniques for carrying out daily duties and activities in occupational therapy.

Behavioral therapy: These behavioral symptoms can be treated with this kind of therapy, along with hyperactivity and sleep disorders.

Speech therapy: A person with Angelman's syndrome can improve their communication skills with the help of speech therapy.

Treatment for scoliosis: Braces or surgery are two options for treating scoliosis, or a curved spine.

Eye surgery: Crossed eyes may be corrected surgically. Angelman syndrome is an uncommon in gastrointestinal condition that can be treated with dietary modifications and drugs which include feeding problems and constipation.

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