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Pediatrics

ANGELMAN SYNDROME

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ABSTRACT Firstly, Harry Angelman was a pediatrician who reported this case. Angelman syndrome is a congenital disorder caused by the absence of a specific gene which is transmitted through mother's chromosome 15 and causes late development, difficulty in speech and balance, intellectual disability, and sometimes, seizures, tremors of limbs, the small size of the head and a matchless behavior with a happy manner that includes recurrent laughing, smiling, and sensibility. The prevalence of Angelman syndrome is approximate to be out of 1,065,070,607 populations, children pretended by Angelman syndrome are 88,755. It shows less prevalence rate. At 12 months of age, the sign or symptoms may start to begin and the most common symptom is delayed development in children. At the age of 2 and 3 years old, Seizures may begin. Angelman syndrome is caused by the loss of the normal contribution to the area of chromosome 15 from the mother which is called the ubiquitin-protein ligase E3A (*UBE3A*) gene. It is equally severe for both males and females. At the time of treatment, health workers focus on leading medical, sleep, and developmental problems. There is no cure for Angelman syndrome but we can treat the child patient on the basis of signs and symptoms. The background of this disease is rare.[1]

KEYWORDS : congenital, gene, development, Angelman syndrome.

1. INTRODUCTION

In 1965, Harry Angelman was a pediatrician working in Warrington, England, firstly she reported in three children with this condition. In India, it equally affects males and females. The prevalence of Angelman syndrome is approximate to be out of 1,065,070,607 populations, children pretended by Angelman syndrome are 88,755. It shows less prevalence rate.[1] Angelman syndrome is a genetic disorder that causes late development, difficulty in speech and balance, intellectual disability, and sometimes, seizures. These types of children with Angelman syndrome generally smile and laugh usually, and have happy, excitable, Or jolly personalities. Mostly in 6-12 months of age, the sign or symptoms may start to begin and the most common symptom is delayed development in children. At the age of 2 and 3 years old, Seizures May begin.[2] People with this disease tend to live a normal lifestyle, but we cannot be cured the disorder. At the time of treatment, we focus on leading medical, sleep, and developmental issues.

2. Definition

It is an uncommon congenital disorder marked as a mental defect and has a tendency to jerky movement and it is also called as "Happy Puppet Syndrome".[2] it caused by the absence of particular genes normally which is present in inherited from the mother's chromosome 15 and it generally involves in the nervous system.[3]

3. Causes

Angelman syndrome is caused by the loss of the normal contribution to the area of chromosome 15 from the mother which is called the ubiquitin-protein ligase E3A (*UBE3A*) gene, most commonly by removal of a section of that chromosome. Some are other causes included:

- Uniparental disomy (UPD),
- Translocation,
- Single gene mutation in that area.

A healthy child receives two copies of chromosome 15 which is getting one from the mother and another from the father. [3]

4. Epidemiology

The risk factors for Angelman syndrome are unknown. In some cases, the finding shows that it occurs due to family history and it may increase the chances of a baby having the disorder but the disease is uncommon, it happens in only 1 of every 10,000 children.

5. Pathophysiology

A mother and father transfer two copies of chromosome 15 to the healthy child. Yet, in the area of the chromosome that is crucial for Angelman syndrome, the maternal and paternal give convey particular genes very differently. This is a result of sex-specific epigenetic form; the biochemical mechanism is DNA methylation. The allele of the gene UBE3A is maternal which is in a normal individual, it is a part of the ubliquitin pathway that is conveyed and the paternal allele is notably calmly in the developing brain. The maternal allele is almost exclusively the active one which situated in the hippocampus and cerebellum if the contribution of the maternal is lost or mutated so in that condition Angelman syndrome is occur.5 (Some other genes on chromosome 15 are maternally marked, and when the paternal tend is lost, by similar mechanisms, the eventuate is Prader–Willi syndrome.) The methylation test that is done for Angelman syndrome (a defect in UBE3A) searches for methylation on the gene's adjoin SNRPN which is calmed by methylation on the maternal replicate of the gene.[5]

While Angelman syndrome can be precipitated by a single variation in the UBE3A gene, the most common genetic deficiency leading to Angelman syndrome is a ~4Mb (megabase) maternal removal in chromosomal region 15q11-13 causing an insufficiency of UBE3A aspect in the paternally imprinted brain areas and after that the sign and symptoms are showing.3

6. Symptoms

Angelman syndrome is having some signs and symptoms and that include: [4]

- Delay in the Developmental period, including no crawling or babbling at 6 to 12 months
- Intellectual defect
- Speechless or minimum of speech
- Difficulty in walking, moving or balancing
- Repeated smiling and laughter
- Happy, volatile personality
- Difficulty in sleeping

Children who have Angelman syndrome may also show the following features:[5]

- Seizures, commonly starting between 2 and 3 years of age
- Rigid or jerky movements
- Small head size, with flatness in the back of the head
- Tongue pushing
- Increased sensitivity to heat
- Ataxia
- Change the color of air, skin, and eyes
- Unusual behaviors, such as hand fluttering and arms moved while walking

7. Diagnosis

- ***History collection*** to collect the family background of parental chromosomal defect.[6]
- ***Physical examination***-for assesses the delayed development of the child and asses the coordination, balance, a small head size, flatness in the back of the head, and recurrent laughter of the child.
- A final diagnosis can almost always be made through a ***blood test Parental DNA pattern-*** This is also known as a DNA methylation test, it is done for identifies three of the four known genetic abnormalities that precipitated Angelman syndrome.
- ***Missing chromosomes-*** A chromosomal microarray (CMA) can do for identifying if any portions of chromosomes are missing.

- ***Gene mutation-*** Hardly, Angelman syndrome may occur when a replicate of the UBE3A gene of a person maternal is active but changed. If the DNA methylation test is normal, your child's doctor may order to does a UBE3A gene sequencing test to check for a maternal mutation.

8. Complications

Complications related to Angelman syndrome contains[7]:

- ***Feeding difficulties-*** Infant may feel Difficulty in coordinating sucking and swallowing.
- ***Hyperactivity*** - In Angelman syndrome, the children move quickly from one activity to another activity, they have less time for attention and they keep the toys and hand inside their mouths.
- ***Sleep disorders-*** The children with Angelman syndrome have frequent abnormal sleeping and waking patterns and they may need less time of sleep than other children
- ***Scoliosis-*** Some of the children with Angelman syndrome develop an unusual side-to-side spinal curvature throughout the time.
- ***Obesity-*** The children of Angelman syndrome tend to have more tendencies to eat the food so that they became obese.

9. Treatment

There is no other specific treatment for Angelman syndrome. Research is concentrating on targeting particular genes for treatment. The present treatment is focusing on handling medical and developmental outcomes. A special health worker's team will appropriately work with you to take care of your child's condition.8 On the basis of the child's sign and symptoms; there are some treatments for Angelman syndrome may include:

- ***Anti-seizure*** medication to control seizures
- ***Physical therapy*** to help to improve with walking and movement problems.[8]
- ***Communication therapy*****,** which may provide to improve sign language and picture communication
- ***Behavior therapy*** is provided to help overcome hyperactivity and a short attention period and to help in development.[9]

10. Prevention

In unusual cases, Angelman syndrome may be crossed defective gene from affected parents. If u already have a same child with the same gene defect so you should talk with your doctor or a genetic counselor for help to make your future planning of other pregnancies.[10]

11. CONCLUSION

Angelman syndrome is an uncommon congenital disorder that is transmitted from parent's chromosome 15 to children which marked as a mental defect and has a tendency to jerky movement. It caused by the absence of particular genes normally. It happens in only 1 of every 10,000 children. Angelman syndrome is having some signs and symptoms and that is a delay in Developmental period, including no crawling or babbling at 6 to 12 months, Intellectual defect, speechless or minimum of speech, difficulty in walking, moving or balancing, repeated smiling and laughter, happy, volatile personality, difficulty in sleeping. It assessed by a blood test, parental DNA pattern, missing chromosomes, and gene mutation test for the assessment of Angelman syndrome. Some complications related to Angelman syndrome contain feeding difficulty, hyperactivity, sleep disorders, curved spinal cord, and obesity. There is no specific treatment for Angelman syndrome but on the basis of child's sign and symptoms that is anti-seizure drugs, physical therapy, communication therapy, and behavior therapy for a treat the Angelman syndrome. There is no cure for Angelman syndrome but we can treat the child patient on the basis of signs and symptoms.

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