Angelman's Syndrome

Angelman’s syndrome is caused by paternal imprinting after the deletion of a gene located on the long arm of chromosome 15. This condition is alternatively known as “Happy Puppet Syndrome” and is characterized by inappropriate laughter, ataxia, seizures, abnormal EEG findings, and intellectual disability.



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Mechanism

Paternal Imprinting

Father Imprinting a Stamp

Imprinted genes are silenced genes, meaning they are not expressed and can be a normal phenomenon. Humans have two sets of genes from their mother and father, and having one set of genes (alleles) imprinted is not an issue in normal cases, as the other gene is present to express the given trait. However, if a mutation occurs on the active allele while the other allele is imprinted, then there will no longer be a functional copy of the gene in that region. In Angelman's syndrome, the paternal allele is imprinted at baseline. The disease results if the maternal allele is mutated or deleted since this results in a lack of a functional gene.

Deletion of Chromosome 15q (Maternal Deletion)

Cutting off Chromosome of (15) Quinceanera-dress and Mother Missing

The affected genes are found on the long arm of chromosome 15, specifically at the locus 15q11-13. Recall that “q” refers to the long arm of a chromosome, while “p” represents the short arm. A mutation on the maternal 15q allele, whether it is a deletion, inversion, etc., will lead to Angelman’s syndrome.

Signs And Symptoms

Happy Puppet Syndrome

Happy Puppet

Although an outdated term, Angelman’s syndrome was once known as “Happy Puppet Syndrome” due to the personality and behavior of these individuals. They smile and laugh constantly and are easily excited.

Inappropriate Laughter

Laughing

Angelman’s patients are known for their unique and seemingly pleasant behavior of laughing and smiling, even if it can be at inappropriate times. They often have short attention spans as well.

Ataxia

A-taxi

A wide-based gait due to poor motor control is characteristic of Angelman’s. Individuals also exhibit hypermotoric behavior with arm waving and hand flapping.

Severe Intellectual Disability (Mental Retardation)

Tar Covered Book

Because of the severe intellectual disability that accompanies Angelman’s syndrome, these patients are unable to live independently and will require lifelong care.

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Seizure

Caesar

Seizures can be a concerning complication of Angelman’s syndrome. Parents should be educated about proper seizure care when caring for affected children.

Abnormal EEG

Abnormal-spikes from the EEG-cap

After making an Angelman's diagnosis, an EEG should be obtained for baseline evaluation. A notable finding with these patients is large-amplitude slow-spike waves, with or without documented seizure activity.

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