Angelman Syndrome

What is Angelman Syndrome?

Angelman syndrome is a rare genetic and neurological disorder characterized by severe developmental delays and learning disabilities; absence or near absence of speech; inability to coordinate voluntary movements and a distinct behavioral pattern characterized by a happy disposition and unprovoked episodes of laughter and smiling. Although those with this syndrome may be unable to speak, many individuals learn to communicate through other means such a gesturing.

What are the symptoms of Angelman Syndrome?

Angelman syndrome is associated with a broad spectrum of possible symptoms. Children with Angelman syndrome experience delays in reaching developmental milestones and have severe learning disabilities. Children with Angelman syndrome also have significant communication difficulties. Most children do not develop the ability to speak more than a few words. Children usually can understand simple commands. Older children and adults may be able to communicate through gesturing and or using communication boards.

Individuals with Angelman syndrome may have microcephaly in which the circumference of the head is smaller than would normally be expected for a child's age and weight. In many cases, epileptic seizures may also occur. Seizures usually begin between one and five years of age and often improve by adolescence.

Adults with Angelman syndrome may have more pronounced facial features such as a more prominent lower jaw (mandibular prognathism). Some individuals may develop abnormal protrusion of the cornea (keratoconus). Mobility may decrease as some individuals grow older and stiffening of the joints (contractures) may also develop. Some older children and adults may be prone to obesity.

How is Angleman Syndrome diagnosed?

A diagnosis of Angelman syndrome may be made based upon a detailed patient history, a thorough clinical evaluation and identification of characteristic findings. About 80% of cases can be confirmed through a variety of specialized blood tests.

How is Angelman Syndrome Treated

Early intervention is important to ensure that children with Angelman syndrome reach their potential. Special services that may be beneficial to children with Angelman syndrome may include special social support and other medical, social, and/or vocational services. Most children with Angelman syndrome benefit from physical, speech and occupational therapy. Behavioral modification therapy may be used to discourage unwanted behaviors.

Resources

- https://rarediseases.org/rare-diseases/angelman-syndrome/
- https://www.angelman.org/resources-education/state-resources/new-jersey/
- http://www.matheny.org/about
- https://ghr.nlm.nih.gov/condition/angelman-syndrome
- https://www.angelman.org/what-is-as/
- http://www.mayoclinic.org/diseases-conditions/angelman-syndrome/basics/definition/con-20033404
- http://www.epilepsy.com/learn/types-epilepsy-syndromes/angelman-syndrome