Fact Sheet :

Angelman Syndrome

WESTERN CAPE ASSOCIATION FOR PERSONS WITH DISABILITIES
TM: WCAPD

What is Angelman Syndrome?

Angelman Syndrome (AS) is a rare neuro-genetic disorder that occurs in 1 in 15,000 live births or 500,000 people worldwide. It is caused by a loss of function of the UBE3A gene in the 15th chromosome derived from the mother, causing delays in brain development.

AS shares symptoms and characteristics with other disorders including autism, cerebral palsy and Prader-Willi syndrome which means that misdiagnosis occurs often. The syndrome was named after Dr Harry Angelman, the doctor who first investigated the symptoms in 1965.

Despite developmental challenges, people with Angelman Syndrome have an overall happy and excitable demeanor and an individual with AS will light up a room with their smile and laughter. Another name for AS is "happy puppet syndrome".

Signs & symptoms

- Developmental delays, incl. no crawling or babbling at 6 to 12 months
- Intellectual disability
- No speech or minimal speech
- Difficulty walking, moving or balancing well
- Frequent smiling and laughter
- Happy, excitable personality
- Trouble going to sleep and staying asleep

Features

- Seizures, usually beginning between 2 & 3 years of age
- Stiff or jerky movements
- Small head size, with flatness in the back of the head
- Tongue thrusting
- Hair, skin and eyes that are light in color
- Unusual behaviors, like hand flapping & arms uplifted while walking

Treatment

There's no cure for Angelman syndrome. Research is focusing on targeting specific genes for treatment. Current treatment focuses on managing the medical and developmental issues. Depending on your child's signs & symptoms, treatment for AS may involve:

- Anti-seizure medication to control seizures
- Physical therapy to help with walking and movement problems
- Communication therapy, which may include sign language and picture communication
- Behavior therapy to help overcome hyperactivity and a short attention span and to aid in development.

Life expectancy & prognosis

Some symptoms of Angelman syndrome improve as individuals get older. Sleep issues and seizures tend to become less severe or infrequent. Because of mobility issues, obesity and scoliosis can develop in adolescence.

The life expectancy of people with Angelman syndrome is normal. AS itself does not cause death. However, there can be severe complications due to some of the symptoms of the syndrome, such as seizures and aspiration pneumonia. There is also the possibility of accidents due to walking and balance issues and attraction to water that can cause severe injury.

Individuals with AS will require life-long care, but can live long, happy lives.

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Resources : *Angelman Syndrome Foundation*
Mayo Clinic