

Angelman Syndrome

Background

This neurodevelopmental disorder, once termed “happy puppet” due to the happy disposition and characteristic movement disorder of the patients, was first described in 1965 by Harry Angelman. Angelman Syndrome is associated with severe intellectual disability, microcephaly (small head), speech impairments, seizures and characteristic behaviour and facial appearance.

Cause of Angelman Syndrome

This disorder is caused by a variety of genetic mechanisms involving chromosome 15. In over 80% of cases clinical diagnosis can be confirmed by laboratory testing. Some genetic mechanisms have implications for recurrence.

The incidence has been estimated as between 1 in 10,000 – 25,000 births.

Presentation

Consistent Characteristics (100% of cases)

- Severe developmental delay.
- Speech impairment with minimal or no use of words. Understanding better than expressive language.
- Movement or balance disorder – often ataxic gait, tremulous movement of hands.
- Behavioural phenotype: frequent laughter, easily excitable, hand flapping, hyperactive, happy demeanour, short attention span.

Frequent Characteristics (80% of cases)

- Absolute or relative microcephaly by the age of 2 years.
- Seizures with onset usually between 1- 3 years.
- Characteristic EEG changes – large amplitude slow-spike waves.

Associated Characteristics (20- 80% of cases)

- Flat occiput, occipital grooves.
- Deep-set eyes.
- Prominent jaw.
- Wide mouth and widely spaced teeth.
- Tongue thrusting, sucking/swallowing disturbance.
- Hypo-pigmented skin, hair and eyes.
- Up-lifted flexed arms when walking.
- Hyperactive lower limb deep tendon reflexes.
- Increased sensitivity to heat.
- Sleep disturbances which improve with age.
- Fascination with water.
- Strabismus.

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Children with Angelman syndrome can acquire some simple skills associated with daily living but none would be able to live independently. By adulthood, about 80% are toilet trained by day.

Diagnosis

Clinical diagnosis is difficult in the first two to three years of life. Diagnostic criteria are based both on clinical features and on the currently available genetic information.

Conditions with a similarities in presentation include Rett syndrome, Lennox Gastaut syndrome, autism and non-specific cerebral palsy.

Developmental History and Investigations

- Normal antenatal and birth history.
- Normal head circumference at birth and absence of major birth defects.
- Developmental delay evident by 6-12 months of age.
- Ongoing delay in development but no regression.
- Normal metabolic, haematological and chemical laboratory profiles.
- No structural brain abnormalities.

Management

Key recommendations for management include:

- Diagnosis and access to a support association
- Genetic counselling
- Management of epilepsy – any type of seizure can occur. Diagnosis and management can be complicated.
- Management of hyperactive behaviour
- Physiotherapy, occupational therapy and speech therapy
- Respite and support for family

For further information contact:

Angelman Syndrome Association Inc
<http://www.angelmansyndrome.org>

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The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

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