

Angelman Syndrome

- to be included in the Personal Health Records of people who have been diagnosed as having Angelman Syndrome

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.

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

CAUSE

This disorder is caused by a variety of genetic mechanisms all involving chromosome 15. In about 80% of cases clinical diagnosis can be confirmed by laboratory testing. Most cases are sporadic but some genetic mechanisms have implications for other family members.

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

DIAGNOSIS

Diagnostic criteria are based both on clinical features and on the currently available genetic information. Clinical diagnosis is difficult in the first two to three years of life. Conditions with similarities in presentation include Rett syndrome, Lennox Gastaut syndrome, autism and non-specific cerebral palsy.

CLINICAL CHARACTERISTICS

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.

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: can include frequent laughter, easily excitable, hand flapping, hyperactive, happy demeanour, short attention span, mouthing objects.

Frequent Characteristics (80% of cases)

- Absolute or relative microcephaly by the age of 2 years.
- Seizures with onset usually before 3 years.
- Characteristic EEG changes – large amplitude slow-wave spikes.

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
- Hypopigmented skin and eyes
- Fair or blond hair
- Hyperactive lower limb deep tendon
- Uplifted flexed arms when walking
- Increased sensitivity to heat
- Sleep disturbances which improve with age
- Strabismus
- Fascination with water

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.

KEY RECOMMENDATIONS FOR MANAGEMENT

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

NATIONAL SUPPORT ASSOCIATION

Angelman Syndrome Association Inc.

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<http://www.angelmansyndrome.org>