

## Angelman syndrome

Angelman syndrome is a genetic condition that is present at birth (congenital). It causes intellectual disability and other features. Other characteristics of Angelman syndrome include distinctive facial features, mental retardation, speech problems and hyperactive behaviour. In most cases, the cause is a missing section (deletion) on the mother's copy of chromosome 15.

This condition was once known as 'happy puppet syndrome' because of the child's sunny outlook and jerky movements. It is now called Angelman syndrome after Harry Angelman, the doctor who first investigated the symptoms in 1965.

Most diagnoses are made between the ages of three and seven years of age. Estimates vary, but Angelman syndrome is thought to affect one child in every 10,000 to 25,000.

### Symptoms that are usually present

Characteristic symptoms of Angelman syndrome that are usually present include:

- Delayed motor development, such as delay in sitting, crawling and walking
- Speech problems
- Jerky, puppet-type movements
- Stiff-legged walking style
- Hand flapping
- Hyperactive behaviour
- Loving, happy and social demeanour
- A child easily moved to laughter
- Intellectual disability – a child with Angelman syndrome will have delayed development in all areas and disability is severe in most cases.

### Symptoms that are sometimes present

Characteristic symptoms of Angelman syndrome that are sometimes present include:

- Small head
- Characteristic EEG (brainwave) abnormalities
- Epilepsy (occurs in 80 per cent of cases).

### Characteristic physical features

The characteristic physical features of this condition are not always obvious at birth but evolve during childhood. Characteristic physical features of Angelman syndrome include:

- Flattened back of the head
- Deep-set eyes
- Wide, ever-smiling mouth
- Prominent jaw and widely spaced teeth
- Lightly pigmented hair, skin and eyes.

### Typical behaviour problems

Some of the common problems include:

- Feeding difficulties
- Disturbed sleep
- Delayed toilet training (about 80 per cent of adults are dry during the day).

### Chromosome disorder

A child inherits two sets of chromosomes – one set from each parent. Angelman syndrome occurs in the following scenarios:

- A section of genetic material is missing from the copy of chromosome 15 inherited from the mother. This is the most common scenario.
- The child inherits two copies of chromosome 15 from its father and none from its mother. This happens occasionally.
- The child may inherit one chromosome 15 from each parent, but the chromosome from the mother works in the same way as the chromosome from the father.
- In 20–30 per cent of cases, there is no cause found. Some of these patients have a fault (mutation) in a gene called UBE3A on chromosome 15.

### **Diagnosis**

Diagnosis methods include checking for the clinical features of Angelman syndrome and performing DNA tests.

Angelman syndrome may be mistaken for autism because of similar symptoms including hyperactive behaviour, speech problems and hand flapping. However, a child with Angelman syndrome is highly sociable, unlike a child with autism. It is important that the child is carefully diagnosed, because sometimes Angelman syndrome and autism are both present.

Other conditions that share some common characteristics with Angelman syndrome include Rett syndrome, Lennox-Gastaut syndrome and non-specific cerebral palsy.

### **Treatment**

There is no cure for Angelman syndrome, but the child can benefit from a range of treatments for some symptoms including:

- Speech therapy
- Behaviour modification
- Communication therapy
- Occupational therapy
- Physical therapy
- Special education
- Social skills training
- Anti-epileptic medication.

Angelman syndrome is not a degenerative disease. Children with Angelman syndrome can expect a normal lifespan.

### **Support for parents**

Support for parents includes:

- Support organisations, such as the Australian Angelman Syndrome Association
- Genetic counselling
- Family therapy
- Respite care.

### **Where to get help**

- Your doctor
- Australian Angelman Syndrome Association [www.angelmansyndrome.org](http://www.angelmansyndrome.org)
- Autism Victoria Tel. (03) 9885 0533
- The Centre for Developmental Disability Health Victoria (CDDHV) Tel. (03) 9564 7511

### **Things to remember**

- Angelman syndrome is a neurological disorder caused by a missing section of chromosome 15.
- Common characteristics include intellectual disability, delayed speech or no speech at all, jerky walking style and happy demeanour.
- There is no cure, but the child can benefit from treatment including physical therapy, special education and behaviour modification.

**This page has been produced in consultation with, and approved by:**

Genetic Health Services Victoria

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