Angelman syndrome

Summary

- Angelman syndrome is a genetic disorder caused by a problem with the UBE3A gene on chromosome 15. People with Angelman syndrome are either missing a copy of that gene, or the copy that they have does not work properly.
- 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.

Angelman syndrome is a genetic condition that is present at birth (congenital).

Most cases occur when a certain gene (the UBE3A gene) on chromosome 15 is missing (deletion). Other causes include the UBE3A gene being incorrectly inactivated or when there is a change (mutation) in this gene.

Characteristics of Angelman syndrome include distinctive facial features, intellectual disability, speech problems, jerky walking style, happy demeanour and hyperactive behaviour.

Angelman syndrome 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 two and five years of age. Estimates vary, but Angelman syndrome is thought to affect one child in every 10,000 to 20,000.

Symptoms of Angelman syndrome

Characteristic symptoms of Angelman syndrome that are usually present include:

- delayed motor development, such as delay in sitting, crawling and walking
- speech problems or no speech at all
- problems with balance and coordination (ataxia)
- jerky, puppet-like 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.

Characteristic symptoms of Angelman syndrome that are sometimes present include:

- small head (microcephaly)
- characteristic brainwave abnormalities
- epilepsy (occurs in more than 80 per cent of cases).

Physical characteristics of Angelman syndrome

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 (microbrachycephaly)
- deep-set eyes
- wide, ever-smiling mouth
- prominent jaw and widely spaced teeth
- lightly pigmented hair, skin and eyes.

Behaviour problems with Angelman syndrome

Some of the common problems include:

- feeding difficulties
- disturbed sleep
- delayed toilet training (about 80 per cent of adults are dry during the day)
- very short attention span.

Causes of Angelman syndrome

A child inherits two sets of chromosomes – one set from each parent. Therefore, children usually inherit one copy of the UBE3A gene on chromosome 15 from each parent. Both of these copies become active in many areas around the body.

In some areas of the brain only the copy of the gene inherited from the mother is active (this is normal). If this gene is missing or there is a change or mutation in it, the person will have no functional copies of the gene in those parts of their brain. This means that they will not have enough UBE3A protein in their brain. This is thought to be what causes Angelman Syndrome.

The main causes of Angelman syndrome can be summarised as follows:

- A section of genetic material (that usually contains the UBE3A gene) is missing from the copy of chromosome 15 inherited from the mother. (This causes ~68 per cent of cases.) Note that the mother's chromosome 15 is normal, and the genetic material is lost during the development of the egg.
- The child has a change (mutation) in the UBE3A gene on chromosome 15, which prevents its expression or function. (This causes ~11 per cent of cases.)
- The child inherits two copies of chromosome 15 from the father. Since the maternal copy is normally needed for proper expression of UBE3A, there will not be normal levels of UBE3A in the brain. (This causes ~7 per cent of cases.)
- The child has a deletion somewhere else on chromosome 15 that leads to loss of expression of the mothers UBE3A gene. (This causes ~3 per cent of cases.)

In approximately 11 per cent of cases, the cause is not known.

Diagnosis of Angelman syndrome

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 for Angelman syndrome

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 of children with Angelman syndrome

Support for parents of children with Angelman syndrome includes:

- support organisations, such as the **Angelman Syndrome Association Australia**
- genetic counselling
- family therapy
- respite care.

The **Genetic Support Network of Victoria (GSNV)** is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by Angelman syndrome.

Genetic counselling and Angelman syndrome

If a family member has been diagnosed with Angelman syndrome, or this genetic condition runs in your family, it can be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand Angelman syndrome and what causes it, how it is inherited, and what a diagnosis means for your child's health and for your family.

Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If Angelman syndrome runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing that condition on to your child, or to arrange for prenatal tests.

Where to get help

- Your **GP (doctor)**
- **Angelman Syndrome Association Australia**
- **Foundation for Angelman Syndrome Therapeutics Australia**
- **Amaze Autism Spectrum Information and Referral Service**Tel. **1300 308 699**
- **Centre for Developmental Disability Health Victoria (CDDHV)** Tel.**(03) 9792 7888**
- **Better Start for Children with Disability** Tel. **1800 242 636**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**

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

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