Saint Mary's Hospital
Manchester Centre for Genomic Medicine

Information for Patients

Angelman Syndrome

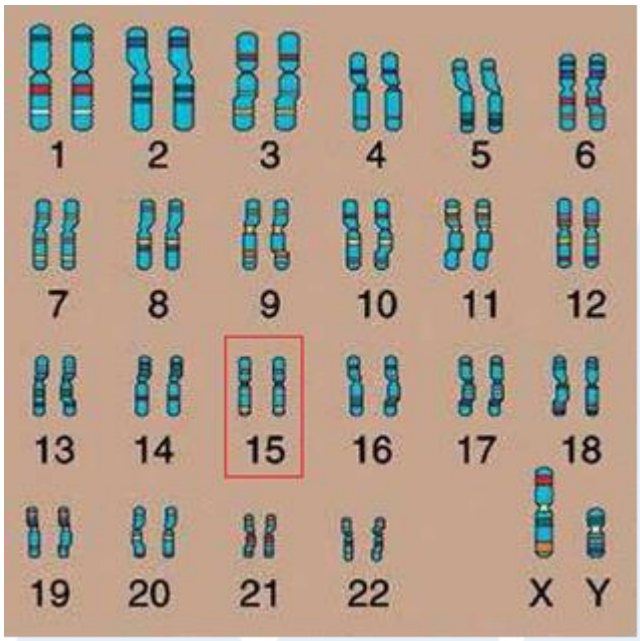
What is Angelman Syndrome?

Angelman Syndrome is a rare developmental disorder that affects 1 person in every 20,000. It is a genetic condition (i.e. it is caused by changes in our genes) which affects parts of the nervous system, particularly the brain. The condition affects both boys and girls.

What are genes and chromosomes?

Our genes are the unique set of instructions inside every cell of our body. Genes determine our personal characteristics such as eye colour and hair colour. There are many thousands of genes, each carrying a different instruction. As well as determining how we look, our genes control the way each cell of the body works. Specific genes control specific cells.

Chromosomes are made up of thousands of genes. These chromosomes are arranged in 23 pairs, according to size as shown in the picture below. We normally inherit one chromosome from each pair from our mother and one from our father.



Why does Angelman Syndrome occur?

Angelman syndrome is caused by a fault in one of the genes on chromosome 15. There are several different ways in which this gene can be affected including:

1. A missing piece of genetic material (deletion) on one of the chromosome 15s. This is the most common cause of Angelman syndrome (75% of cases).

2. An alteration (similar to a spelling mistake) in the Angelman gene UBE3A (10% of cases).

3. Both copies of chromosome 15 are inherited from the father (3% of cases) instead of the normal situation where we usually inherit one chromosome 15 from our mother and one from our father. This is known as uniparental disomy.

4. Inheriting a 'switched off' copy of the Angelman gene from one's mother (2%). This is known as imprinting defect.

5. A chromosome rearrangement is present which affects the way the Angelman gene is working.

In a small number of cases we are unable to find a genetic cause for Angelman syndrome.
In these individuals tests for other Angelman-like disorders are recommended.

Early symptoms of Angelman syndrome may include:

- Feeding problems.
- Delayed milestones.
- Unusual movements including fine tremors and jerky limb movements.
- Epilepsy with an abnormal EEG pattern.
- Lack of speech development and baby babble.
- Poor sleep patterns.
- Happy demeanour.
- Hand flapping particularly when excited.
- Low muscle tone.

As the children grow they may show:

- A flattened back of head.
- Light skin and hair colour (compared to other family members).
- A tendency to chew objects.
- Difficulties with learning, especially affecting speech.
- A similar facial appearance to other children with Angelman syndrome.
- A tendency to laugh even for very minor reasons.
- An increased chance of developing curvature of the spine.

All patients with Angelman syndrome will benefit from occupational therapy, physiotherapy and speech and language therapy.

Whilst most individuals with Angelman Syndrome are diagnosed in childhood, the diagnosis should also be considered in adults with learning disabilities who show the same characteristic pattern of behaviour. This is because doctors are getting better at recognising it and diagnostic techniques have improved. Many individuals with Angelman syndrome have near-normal life expectancy.

Positive aspects

Most people with Angelman syndrome are sociable, loving and generally happy.

They tend to love water, noisy toys, balloons, balls, TV and music.

Some show considerable understanding of verbal and non-verbal communication.

Some learn sign language and use communication devices effectively.

Seizures improve with age during childhood.

A few children develop basic speech.

Significant progress can be made following early intervention.

Learning continues throughout life.

Medical research is beginning to identify new treatments which are being tested in clinical trials.

Is it possible to have a second child with Angelman Syndrome?

If you have a child with Angelman syndrome, in most cases, the risk of having a second affected child is low (about 1%).

However, the risk may be higher than this in some families. The risk depends on the type of genetic alteration your child has and your consultant geneticist will be able to tell you the risk for your family once this has been established.

For more information

If you need more advice about any aspect of Angelman Syndrome, you are welcome to contact:

Manchester Centre for Genomic Medicine
Sixth Floor
Saint Mary's Hospital
Oxford Road Manchester M13 9WL

Telephone: (0161) 276 6506 (Reception)
Facsimile: (0161) 276 6145

Department staffed Monday–Friday, 9.00 am to 5.00 pm.

Website: www.mangen.co.uk

There is a support group that provides information packs and has a parent helpline:

ASSERT
Angelman UK
PO BOX 4692
Nuneaton CV11 9FD

Parent Helpline: 0300 999 0102
Registered Charity Number: 1021882

Please let us know if you would like this leaflet in another format (e.g. large print, Braille, audio).

We would like to acknowledge our Clinical Genetics colleagues at Guy's and St Thomas' Hospital NHS Trust who originally designed and wrote the first version of this leaflet.

Seen in clinic by (doctor): _____

And (Genetic Counsellor): _____

Telephone number: _____

Family reference number: _____