

For more information:

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

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

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

A.S.S.E.R.T.

PO Box 505

Sittingbourne

Kent

ME10 1BR

Parents Helpline: 01980 652617

Registered Charity Number: 1021882

This leaflet was written by Clinical Genetics, Guys Hospital. Updated by Genetic Interest Group Scotland. Last updated Jan 2002.

Seen in clinic by.....

Angelman Syndrome



General information

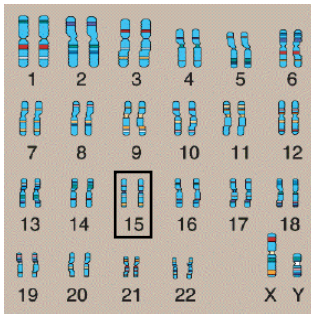
What is Angelman syndrome?

Angelman Syndrome is a rare condition 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 thousand of genes, each carrying a different instruction. As well as determining how we look, our genes control the way each cell or building block 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 of each of the pairs 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:

- A) 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).
 - B) An alteration (similar to a spelling mistake) in the Angelman gene (10% of cases).
 - C) Both copies of chromosome 15 are inherited from ones father (3% of cases). We usually inherit one chromosome 15 from our mother and one from our father.
 - D) Inheriting a "switched off" copy of the Angelman gene from ones mother (2%).
 - E) A chromosome rearrangement.
- In 10% of cases we are unable to find a genetic cause for Angelman syndrome.

Early symptoms of Angelman syndrome may include:

- Feeding problems.
- Delayed milestones.
- Unusual movements including fine tremors and jerky limb movements.
- Epilepsy.
- 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.
- Severe learning and physical difficulties.
- A similar facial appearance to other children with Angelman syndrome
- A tendency to laugh a great deal.

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

Recently the diagnosis of Angelman syndrome is being made more frequently in adults with disabilities. This is because doctors are getting better at recognising it and diagnostic techniques have improved. A person with Angelman syndrome has a 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.
- Sleeping improves with age.
- Seizures improve with age.
- Some children develop basic speech.
- Significant progress can be made by early intervention.
- Learning continues throughout life.
- Medical research may improve therapies in years to come.

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 a few 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.