If you are reading this you may have received a confirmed or suspected diagnosis of Angelman Syndrome for your child.

It is likely that you have never heard the words Angelman Syndrome and will be learning for the first time how it affects your child.

You may be feeling afraid, confused, distressed, or overwhelmed. We offer information that can help.

We want to let you know that it does get better – we know, because we have been there too.

This leaflet is intended as an initial source of information with some advice on where to get more.

Further information about Angelman Syndrome and how we can support you is available as follows:

WEBSITE: www.angelman.ie

EMAIL: admin@angelman.ie

Angelman Syndrome Ireland

AngelmanIRE

Angelman Syndrome

There are also many excellent international resources for information that you can access such as:

ASSERT
www.angelmanuk.org

Angelman Syndrome Foundation
www.angelman.org

FAST
www.cureangelman.org

Angelman Syndrome Association
www.angelmansyndrome.org

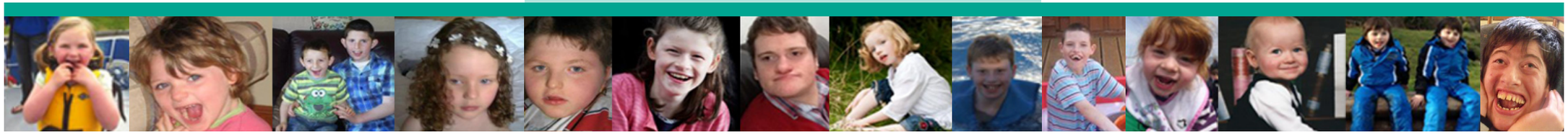
Disclaimer & References
The information contained in this leaflet should not be taken as a substitute for professional medical advice.

The information presented here has been reviewed by a qualified paediatrician, geneticist and neurologist with experience in Angelman Syndrome prior to print.

Angelman Syndrome Ireland

A Guide For Parents

Revision 001 (original) - March 2012



What is Angelman Syndrome?

As part of our DNA, we each have 23 pairs of chromosomes, with one of each pair coming from the mother and the other from the father. Angelman Syndrome (or AS) is a disorder of Chromosome 15.

There are several ways in which Chromosome 15 may be affected. Your medical professional will explain which one applies to your child.

Why did it happen?

The chromosome disorder was there when your child was conceived and there is nothing you or your partner could have done to have caused or prevented it from happening.

In about 95% of cases the cause is a random occurrence with no family history. For subsequent pregnancies the risk of recurrence is very little different to the general population.

In about 5% of cases the disorder is inherited. The genetic situation is different for each famiy and if planning further children then you should discuss your own individual situation with your local geneticist.

How common is it?

Angelman Syndrome affects about **1 in 20,000 - 25,000** individuals. **That means there are roughly 250-350 people in Ireland with AS.** It affects males & females equally and is present across all races and nationalities.

What are the features?*

Below are a collection of symptoms seen in individuals with Angelman Syndrome. AS is a disorder, not a disease and that means affected individuals can live healthy, happy lives. This disorder does, however, have symptoms that should be monitored and managed. Not all of the problems listed below occur in all people. It is not possible to know how many of these features your child might have or to what extent they will be affected by them. There may be many problems or there may only be a few and these problems may be very mild or may be more severe.

Consistent

Developmental delay, functionally severe
Speech impairment, no or minimal use of words
Movement or balance disorder
Apparent happy demeanour; easily excitable personality, often with hand flapping movements; short attention span

Frequent

Seizures, onset usually before 3 years of age
Tongue thrusting or protruding;
Suck or swallowing disorder; drooling
Feeding problems during infancy
Wide mouth, wide-spaced teeth
Excessive chewing/mouthing behaviours
Light skin, hair and eye colour (compared to family)
Increased sensitivity to heat
Sleep disturbance
Attraction to or fascination with water

** Please refer to our website for the full list of clinical characteristics*

Types of Angelman Syndrome Explained:

75%

Deletion
The majority of AS cases are caused by deletions on the maternal copy of Chromosome 15. This deletion prevents the normal expression of a gene called UBE3A in individuals with AS.

10%

UBE3A mutation
Mutations in the UBE3A gene either prevent its expression or function. Thus these individuals do not have the appropriate levels of functional UBE3A in the brain.

7%

Uniparental Disomy
In UPD, the individual has two copies of paternal Chromosome 15. Because UBE3A is not expressed from the paternal copy, these individuals lack normal levels of UBE3A in the brain.

5%

Clinical/Other
In these individuals, all testing for Angelman Syndrome is normal, but they still meet the diagnostic criteria for AS. Please note that there are several other syndromes that present like AS that can be tested for.

3%

Imprinting defect
These individuals may have a deletion of the imprinting centre an Chromosome 15. Loss of imprinting will prevent expression of the maternal UBE3A gene in the brain.

What happens now?

It is likely that your child's blood has been taken to test for Angelman Syndrome and confirm the diagnosis. You will have to wait several weeks for the results. At some point you may be offered Genetic counselling.

To ensure your child has the chance to reach their full potential, you will be referred to the local health provider's "Early Intervention Team". This is a group of professionals who can help your child with advice on Physiotherapy, Occupational Therapy, Epilepsy, Communication skills and other aspects important to your child's development. There may be a waiting list before your child can be seen.

What treatments are available?

In addition to developmental interventions there are medical treatments available to help your child with epilepsy, nutrition, sleep and many other aspects. Your medical professional will assess your child's needs and make appropriate recommendations.

There is an ongoing programme of research and clinical trials around the world to identify a treatment for AS to assist with cognitive skills, motor function and epileptic activity. Some promising advances have been made. More information is available on our website.

What help is available?

Your medical professional can help you with questions you have about the diagnosis or can direct you to health professionals with specific expertise.

Angelman Syndrome Ireland was set up as an All Ireland Charity to help support parents, caregivers and service providers. We have a wide network of parents all over the country and can put you in touch with a parent in your area if you wish to talk about the diagnosis. We have an active online forum for parent-to-parent discussions and support and we also host family information days and an annual conference.

This community may be able to help you identify strategies and techniques such as communication, behavioural or occupational therapies which help with the day to day living with Angelman Syndrome for both the person and family members

Angelman Syndrome Ireland

Angelman Syndrome Ireland
AngelmanIRE
www.angelman.ie