THE ANGELMAN NETWORK

Connecting and supporting those affected by Angelman Syndrome (AS)

Te Hononga Angelman, kia whai hononga me te tautoko hoki i a rátou e pá kii ana ki te mate Angelman Syndrome (AS)

About The Angelman Network (TAN)

The Angelman Network was established as a registered Charitable Trust in New Zealand (CC46746), in 2011. We are the only organisation in NZ dedicated to providing support and up-to-date information on Angelman Syndrome.

Given that families were often struggling to keep up with day-to-day challenges and have very little spare time to search for resources, supports and tested advice, our vision was to collate the latest information from reliable sources on a website that would be regularly updated. This could help to provide immediate access to a wide range of relevant information for families and professionals worldwide.

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www.angelman.org.nz

Our Mission

1. To network nationally and internationally with organizations, specialists, families/whānau and carers in order to access and distribute accurate and up-to-date information about AS.
2. To fully support the 21st century values of inclusion and neurodiversity as specified in the Convention on the Rights of Persons with Disabilities; signed by NZ in 2007 and ratified in 2008.
3. To advocate for the Right to a Voice and to support and promote the global Communication Revolution for non-speakers using the new language of Augmentative and Alternative Communication (AAC) made available by advances in technology.
4. To support and advocate for families so they can empower those with AS to realize their potential and aspirations through person-centred planning.
5. To promote global collaboration for AS research and support the Global Angelman Syndrome Registry.

We also wish to connect NZ families/whānau so they feel supported and valued; to raise more awareness of Angelman Syndrome in New Zealand - among clinicians, therapists, schools, agencies and the general public; and to pursue opportunities to support Angelman Syndrome research in NZ and around the world.

Our Vision / O motor kitenga

That people with Angelman syndrome are / Te tangata mau i te mate AS ka:

- accepted and acknowledged as unique individuals / whakaaetia, manakotia hoki kia u ki to rátou mana ake
- given access to a full language system (AAC) so that their voices can be heard / kia whiwhi ai ki nga ara reo kia rongohia ai o ratou reo e te katoa ma roto i te àpititanga me etahi atu whakamòhiotanga
- valued as contributing members of their community / kia whai hua rátou hei tángata tákoha nó tó rátou hápori

Get in Touch

We invite families, professionals and supporters to become members of The Angelman Network Trust organisation, to support our work. We keep members updated with regular newsletters and notifications about all things Angelman-related. We are always looking out for new members who might be interested in actively supporting our cause by becoming a trustee.

If you have any questions or would like to get in touch, email us at: angelmannetwork@gmail.com

Find us on Social Media

Please 'Like' our official FB page to keep in touch with the latest news articles and updates.

- The Angelman Network (Facebook page)

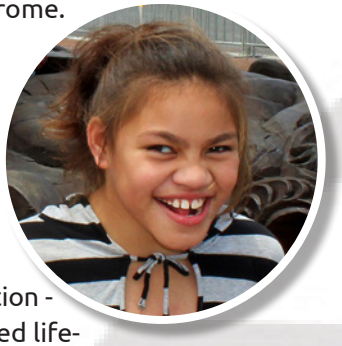
Join our Facebook Groups:

- The Angelman Network (international group)
- Angelman Syndrome NZ (closed group for Kiwis)
- NZ Angel-mums (closed group for mums/primary caregivers)

FACT SHEET – ANGELMAN SYNDROME

What is Angelman syndrome (AS)?

Angelman syndrome is a genetic disorder caused by abnormal function of the gene UBE3A, located within a small region (q11-q13) on the 15th chromosome. A 'syndrome' means a group of recognisable characteristics occurring together. In the 1960s, English paediatrician, Dr Harry Angelman recognised a common set of unusual characteristics in 3 children. He published a medical paper in 1965 documenting his observations. It was only in the early 1990s that genetic testing was developed for what is now known as Angelman syndrome.



- AS is a rare condition that occurs in approximately 1:15,000
- AS affects males and females of all racial/ethnic groups equally
- AS is not a degenerative condition - with a healthy and well managed life-style, individuals with AS have a normal life expectancy but will require life-long care

Angelman Syndrome Characteristics

The syndrome causes delays in development and learning, and a severe speech impairment (many have no words at all).

- Communication occurs naturally by employing more than one mode of communication, eg. using sounds, gestures, posturing, and eye direction.
- In infancy and early childhood, many have feeding complications and appear to require very little sleep.
- Epilepsy occurs in approx. 80% of cases, usually beginning at around 2 years of age.
- Most tend to be very happy and social. At times the smiles and laughter can also express other feelings such as anxiety, fear, and even pain.
- When young, most have a component of hyperactivity. Attention span can be so short that social interaction and communication is adversely affected. This decreases with age.
- A strong attraction for water, music, lights and reflections can be common early on and water can therefore pose a potential accident risk.
- A movement/balance disorder (ataxia) that results in an unusual gait and jerky movements.

Everyone is Unique

It is important for professionals to note that each person who has AS is an individual. Though they have AS, they will also closely resemble their parents/family and will have their own unique appearance and personality. Just like other children, they will also vary greatly in their abilities, strengths and weaknesses. Because of these differences, treatments and management will vary from case to case.

Communication Support

Though people with AS have complex communication needs, their receptive language is typically more advanced than their expressive language. This makes them excellent candidates for Augmentative and Alternative Communication (AAC). Immediately after diagnoses, families should be advised to access AAC support through Talklink Trust.

What causes Angelman Syndrome?

AS occurs through 4 different genetic mechanisms (genotypes) all involving chromosome 15. In about 80% of cases, clinical diagnosis can be confirmed by laboratory testing. Most cases are sporadic but some genetic mechanisms have implications for other family members.

1. Deletion+ (15q11.2-q13 deletions), 68% of cases are 'deletion positive' - The majority of AS cases are caused by deletions on the maternal copy of Chromosome 15.

2. UBE3A Mutations, 11% of cases – 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.

3. Uniparental Disomy, UPD; 7% of cases –The individual has two copies of paternal Chromosome 15.

4. Imprinting Defect, ICD, 3% of cases – A deletion of the imprinting centre on Chromosome 15. Could also be caused by loss of imprinting information during the mother's oogenesis.

5. Clinical/other, 11% – all testing for Angelman Syndrome is negative but they still meet the diagnostic criteria for AS. These individuals may have as yet unrecognized mutations that affect UBE3A or genomic imprinting on Chromosome 15.