

YOU ARE NOT ALONE.

There is a whole community in Australia working together to support families with Angelman syndrome.

Angelman Syndrome Association Australia (ASAA) provides support, information, education, and networking to help navigate life with Angelman syndrome.

The future looks bright. The Foundation for Angelman Syndrome Therapeutics (FAST) funds research to provide treatments to improve the symptoms of AS and ultimately to provide a cure.

Join our support network:
tiny.cc/angelmanaustralia



GLOBAL
ANGELMAN
SYNDROME
REGISTRY

The Global Angelman Syndrome Registry is a tool to help medical professionals and researchers learn more about individuals with AS. The Registry is creating opportunities, understanding and insight about AS, providing an important tool for both facilitating research and enabling clinical trial sponsors to quickly identify suitable patients for each study.

Your participation is vital. Join today.
Angelmanregistry.info



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LIVING WITH ANGELMAN SYNDROME

Cover image supplied courtesy of Lana Pratt Photography





ABOUT ANGELMAN SYNDROME

Angelman syndrome (AS) is a rare genetic disorder that affects approximately 1 in 15,000 live births*.

Symptoms include: global developmental delay, impaired movement and balance, lack of speech, seizures, feeding and sleep difficulties. Early diagnosis is critical; however, AS is often misdiagnosed as cerebral palsy or autism. People living with AS require life-long care and close medical supervision. Intensive educational and therapeutic programs significantly assist skill development and inclusion.

Despite the challenges, individuals with AS are renowned for their happy disposition and huge smiles. Their infectious laughter and emotions are expressed with every inch of their body, with hugs so sincere that they are rarely forgotten! However, their happy, loving nature is often overshadowed by the overwhelming difficulties they, and their carers, face daily. Angelman syndrome is not life limiting or degenerative and individuals are lifelong learners.

WHAT CAUSES AS?

Angelman syndrome is caused by the lack of just one functional gene – UBE3A on chromosome 15. This gene is responsible for creating a protein (of the same name) that is expressed in the brain. There are five known mechanisms – a Chromosome Deletion, Mutation, Imprinting Centre Defect, Uniparental Disomy, Mosaic.

HOW IS IT DIAGNOSED?

New, more widely available DNA microarray testing has made it easier to test for Angelman syndrome. If you suspect your loved one has AS, your doctor or paediatrician can arrange for the test. The array will detect a deletion on Chromosome 15, the most common cause for AS however approximately 30% of cases will require further testing to confirm or rule out Angelman syndrome.

IS THERE A CURE?

Researchers are working hard to deliver therapeutics to improve and ultimately alleviate all symptoms of AS and there is a shared belief in the research community that this is possible.

Research into AS has advanced rapidly in the last ten years and there are a number of gene therapy approaches and treatment options in development. To learn more about these developments visit cureangelman.org.au



WHAT SUPPORT IS AVAILABLE TO ME?

We are fortunate in Australia to have two dedicated groups working together to provide support to the Angelman community. Angelman Syndrome Association Australia (ASAA) is focussed on providing family support for all the day to day aspects of managing and planning with Angelman Syndrome, the Foundation for Angelman Syndrome Therapeutics Australia (FAST) is focussed on funding research and initiatives that lead towards treatments. Both organisations work collaboratively to advocate for our loved ones.

The goal of ASAA is to support, inform, educate, network, promote research and to advocate for families affected by Angelman syndrome. People with AS are eligible for the NDIS and many other State and Commonwealth supports – you can talk to the ASAA on how to access these supports.

The goal of FAST is to fund and collaborate in research aimed at improving the quality of life and understanding for individuals with Angelman syndrome. When new information is made publically available, FAST will help to distribute and educate on the developments including where hard-earned donations are spent. FAST can help you to raise funds in your local community or on a large scale to help further research into Angelman syndrome.



FAST
Foundation for Angelman
Syndrome Therapeutics
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ASA
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