what is angelman syndrome?

angelman uk
support education research

Angelman Syndrome (AS) is a genetic condition, identified in 1965 by British doctor, Harry Angelman. Advances in genetic medicine have made it possible to diagnose increasing numbers of cases and identify the cause. AS cannot currently be cured however science is making huge advances towards possible future treatments.

Angelman Syndrome is a rare neurological disorder affecting 1:15- 20,000 people. It is caused by irregularities on the maternal chromosome 15. (gene UBE3A). People with AS have a normal life expectancy but will require lifelong care and support. Most people with AS are diagnosed between the ages of 3 - 7 yrs old when characteristic features become evident.

primary characteristics

(not conclusive)

- Global developmental delay.
- Complex Communication Needs. Minimal or no use of spoken words. Use of Augmentative & Alternative Communication (AAC) is encouraged for people of all ages. No one is too young or too old to start!
- Sleep disorder. People with AS can remain awake for long periods of time resulting in sleep deprivation for the whole family.
- Epilepsy/Seizures. People with AS can have multiple seizure types including refractory epilepsy. Seizure activity can be worse when young.
- Movement or balance disorder, usually ataxia of gait and/or tremulous movement of limbs.
- Behavioural uniqueness: any combination of frequent laughter/smiling; apparent happy demeanour; easily excitable personality, often with hand flapping movements; hypermotoric behaviour; short attention span.
- Microcephaly.
- Scoliosis.
- Strabismus.
- Cortical Myoclonus/tremor.
- Fascination with & attraction to water.
- Increased sensitivity to heat.
- GI issues/constipation.
- Feeding problems.
- For more information please see our website.

People with Angelman Syndrome are highly sociable & friendly. They are very determined individuals who thrive on positive social interaction. They have wonderful personalities, big smiles and strong hugs!

Whilst a diagnosis of Angelman Syndrome is life changing for all involved, it brings with it connection to an international support network and a local community of families who understand the issues and complexities of living with a rare genetic disorder.

Please turn over to see how AngelmanUK supports UK families and professionals. You can contact us using the details below.

AngelmanUK PO Box 4962 Nuneaton CV11 9FD 0300 999 0102 info@angelmanuk.org www.angelmanuk.org

hello

angelman UK
support education research

AngelmanUK is a registered charity supporting UK families living with Angelman Syndrome (AS). People with AS have have wonderful personalities, big smiles and strong hugs! They also have high support needs, learning disabilities, epilepsy, complex communication needs and associated physical disabilities.

Turn over for more info or see our website.

what we do

- Raise awareness of Angelman Syndrome and encourage mutual support.
- Maintain a 24-hour, 7-days-a-week telephone support line, manned by parents and family members.
- Host a website, social media pages and produce a free newsletter dedicated to Angelman Syndrome.
- Hold regional meetings, a biennial conference, educational workshops and social events.
- Support local research into AS for the benefit of our families and the community.
- Forge links and work closely with international organisations, pooling information and resources.
- Work in collaboration with the Angelman Syndrome Alliance (angelmanalliance.org) pooling finances to fund international scientific research.
- Answer practical queries from medical and social care professionals.

we'd love to meet you

The diagnosis of a rare disorder can be accompanied by feelings of isolation or bewilderment in parents and family members.

Our biennial national conference offers families access to information, education, research studies, fun excursions and entertainment. It is heavily subsidised by AngelmanUK who welcome all who wish to attend and we actively encourage the whole family to come, including siblings of all ages.

thank you!

Registered Charity 1021882

AngelmanUK PO Box 4962 Nuneaton CV11 9FD 0300 999 0102 info@angelmanuk.org www.angelmanuk.org