What is Angelman syndrome?

Angelman Syndrome
is a genetic disorder of Chromosome 15 affecting a gene called UBE3A resulting in a severe developmental delay

The condition gets it's name from the doctor who first described it in 1965 – **Dr Harry Angelman**

What are the features?

Developmental Delay
No or minimal use of words
Movement disorder, Seizures
Sleep Disturbance
Light hair & skin

and remember...

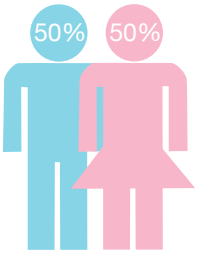
People with AS have great smiles & warm personalities

AS is a Disorder not a disease so individuals can live happy & healthy lives when symptoms such as epilepsy are under control

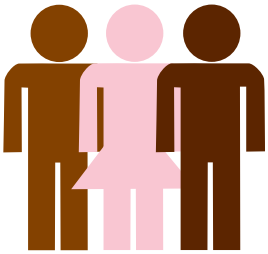
How common is it?

18 people worldwide are born with AS every day

Who does it affect?



It affects males & female equally



It is present in all nationalities, races and ethnic groups

Why did it happen?

Chromosome disorder occurred at the point of conception and nothing the mother or father did could have caused it or prevented it from happening

5% cases are inherited

95% cases are completely random

Scientists are researching ways to treat the cognitive, motor and epileptic aspects of AS

Can it be treated?

People with AS benefit from therapies such as Communication, Physiotherapy and Occupational therapy. There are medical treatments to help with epilepsy, nutrition and sleep

Timeline of Angelman Syndrome:

1965
Dr Harry Angelman publishes a report entitled "Puppet Children" about 3 children he observed

1980's
Research into AS begins in University of Florida under the direction of Dr Charles Williams

1982
Name changed from 'Happy Puppet' to Angelman Syndrome

1987
Discovery that absent genetic code on maternal Chomosome 15 is the genetic marker for AS

1997
The cause of AS discovered by Dr. Joseph Wagstaff & Dr. Arthur Beaudet – mutation or deletion of UBE3A gene

2007
Neurological deficits can be reversed in a mouse model with AS

2011
Dr Ben Philpot discovers how to "switch on" the silent paternal genetic code on Chromosome 15 in a mouse

2012
Dr Ed Weeber begins clinical trials with 24 children using Minocycline to treat cognition and motor function

The Future
HOPE IS IN OUR GENES!