In 1965, Dr. Harry Angelman first described the characteristics that later became known as Angelman Syndrome.

It was considered to be extremely rare, until the 1980s when North American reports began to appear.

Now it is estimated between 1 in 12,000 to 20,000 people.
WHAT CAUSES ANGELMAN SYNDROME

**Loss of function of a gene called UBE3A.**

- **Deletion** - 70%
  - a segment of the maternal chromosome 15 is deleted.

- **Mutation** - 11%
  - in the maternal copy of UBE3A.

- **Paternal Uniparental Disomy** - Small number
  - 2 paternal copies of chromosome 15 are inherited

- **Translocation** - Rarely occurs
  - a mutation or other defect occurs in the area of DNA responsible for activation of the UBE3A.

- The causes in 10-15% of the cases are unknown.
Symptomatic treatments involve:

- Antiepileptic Drugs (seizures)
- Stimulants (hyperactivity)
- Diphenylhydramines (nighttime wakefulness)
- Thorco-lumbar jackets (scoliosis)
- Surgery (orthopedic and other physical problems)
- Educational Training
- Physical Therapy

There are currently no preventative treatments for Angelman Syndrome.
Clinical trials are attempting to augment DNA methylation pathways and increase the expression of the paternal UBE3A, however, initial trials did not show significant clinical benefit.
A possible breakthrough using topoisomerase inhibitors to un-silence the UBE3A gene.

The research examined more than 2,000 drugs to un-silence the paternal gene in mice.
REFERENCES