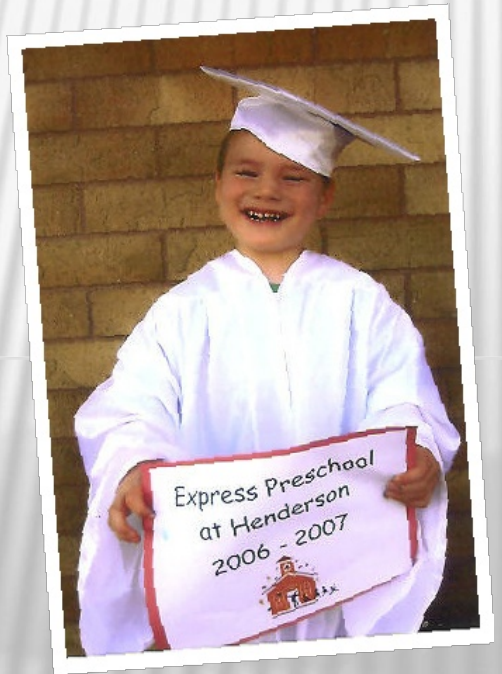


ANGELMAN SYNDROME

Vanessa Gallegos

ANGELMAN SYNDROME

CLASSICAL DIAGNOSIS

- 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

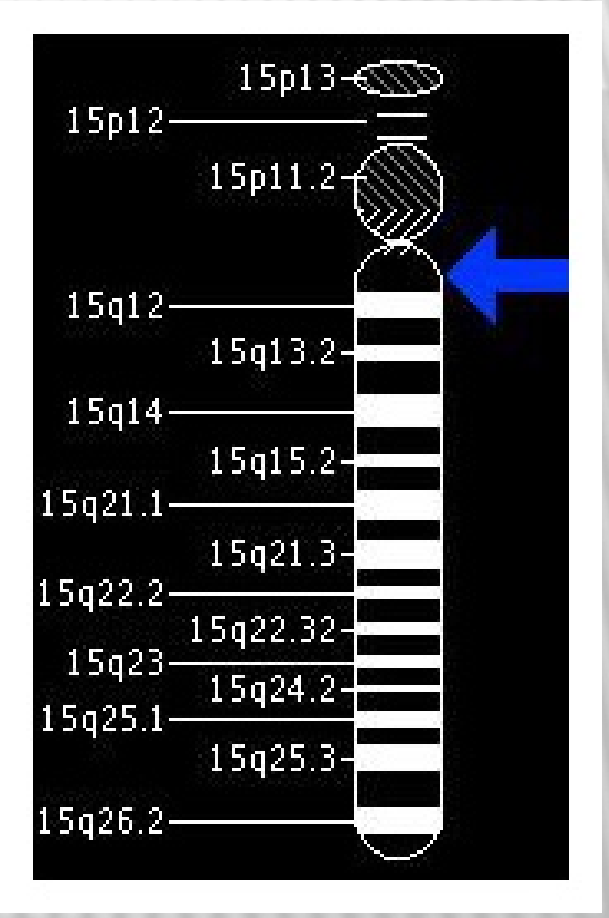
- Loss of function of a gene called *UBE3A*.
  - **Deletion-** 70%
    - a segment of the maternal chromosome 15 is deleted.
  - **Mutation-** 11%
    - in the maternal copy of *UBE3A.*
  - **Paternal Uniparental Disomy-** Small number
    - 2 paternal copies of chromosome 15 are inherited
  - **Translocation-** 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.

TREATMENT

- 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.

THE TRIALS

- 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.



THE FUTURE

- 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.

ANGELMAN
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REFERENCES

- http://www.angelman.org/stay-informed/facts-about-angelma
- http://beaconnews.suntimes.com/news/9935048-418/aurora
- http://ghr.nlm.nih.gov/condition/angelman-syndrome