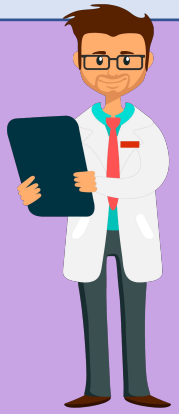


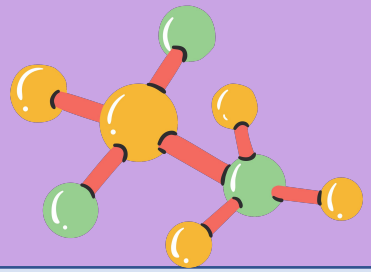


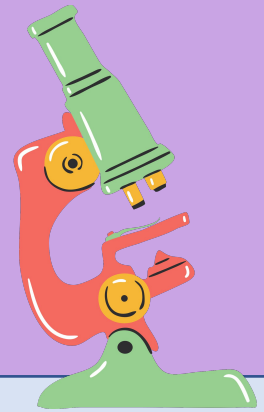




Angelman Syndrome

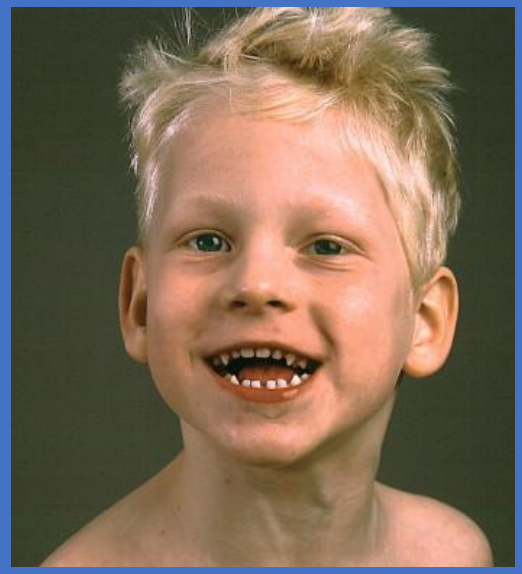
Teacher – Svetlana Smirnova

Presented by-Gracy Singh Dhauni

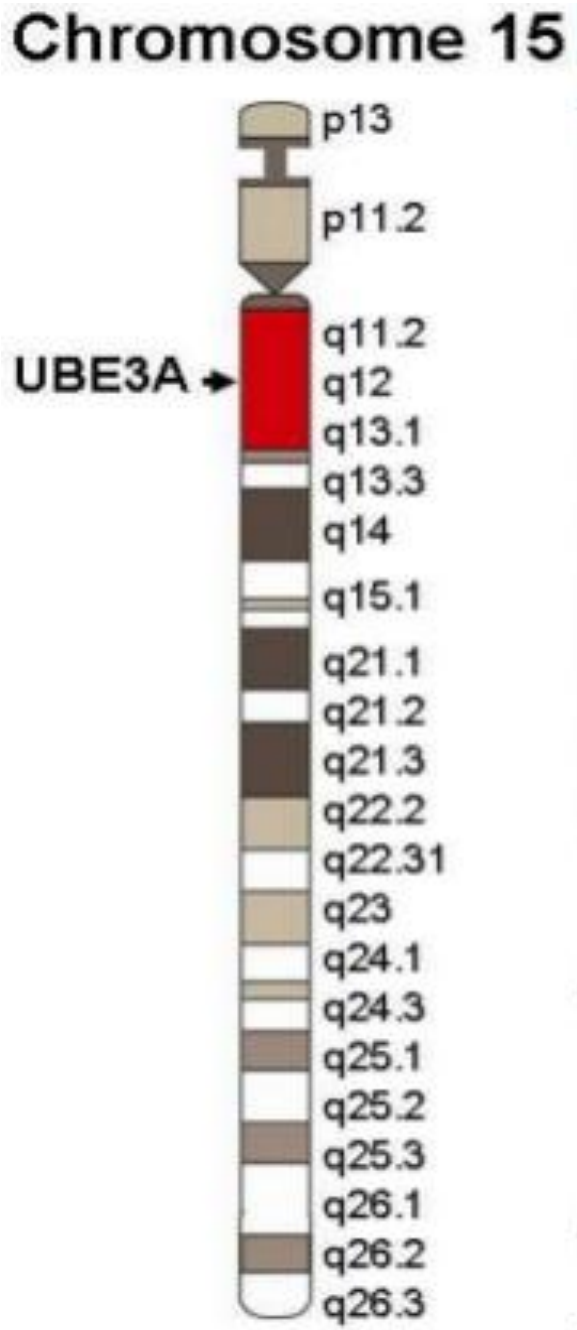



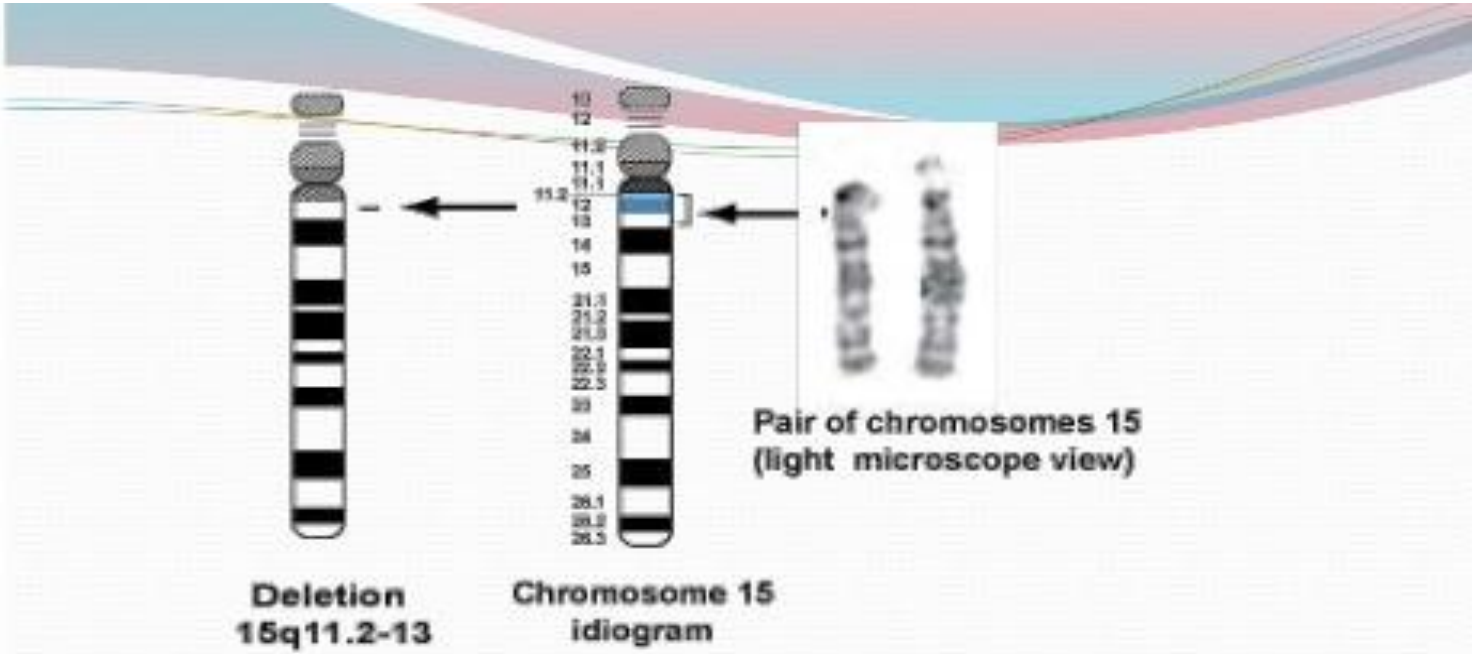



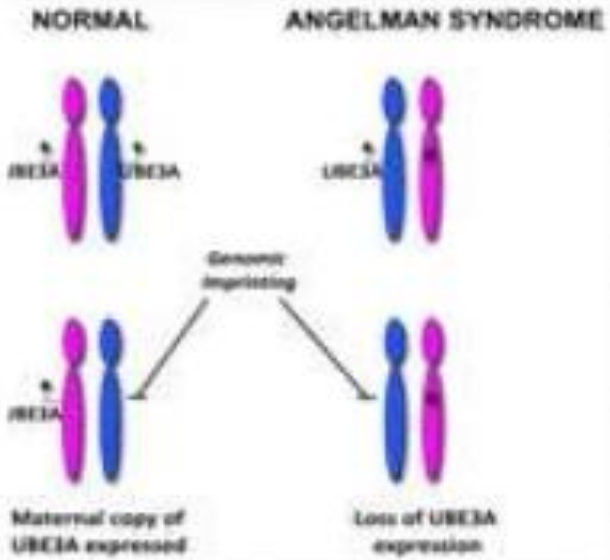
Angelman Syndrome

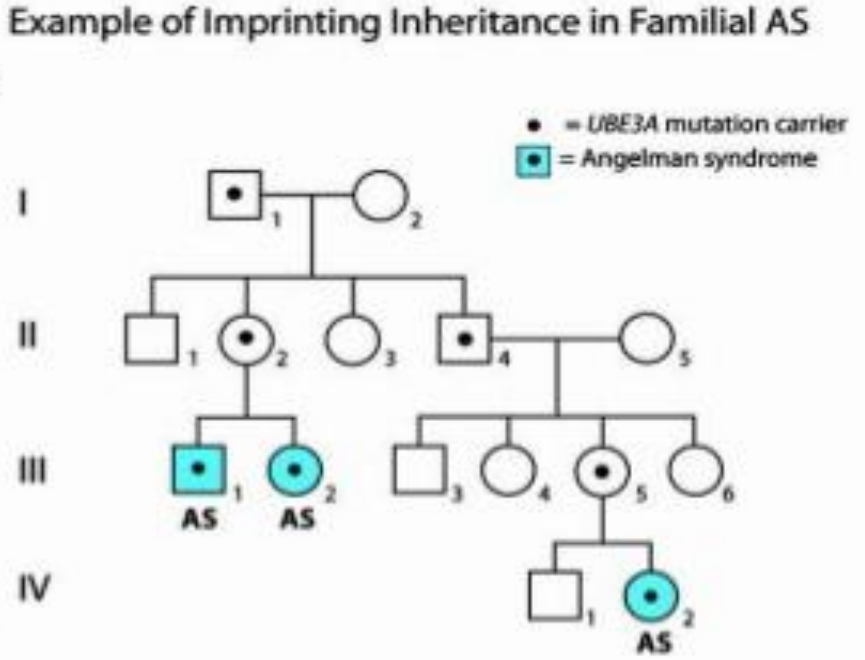


- Angelman syndrome is a genetic disorder that affects 1 out of 15,000 people.. It causes delayed development, problems with speech and balance, intellectual disability, and sometimes, seizures. This disorder affects the nervous system.
- It is resulting from a defect in the maternally inherited copy of chromosomes 15.
- Meaning that chromosome 15 is missing, and there is a break in part of chromosome 11-13.
- People with Angelman syndrome often smile and laugh frequently, and have happy, excitable personalities.
- Developmental delays, which begin between about 6 and 12 months of age, are usually the first signs of Angelman syndrome. Seizures may begin between the ages of 2 and 3 years old.
- People with Angelman syndrome tend to live close to a normal life span, but the disorder can't be cured. Treatment focuses on managing medical, sleep and developmental issues.


Chromosome 15
p13
p11.2
UBE3A
q11.2
q12
q13.1
q13.3
q14
q15.1
q21.1
q21.2
q21.3
q22.2
q22.31
q23
q24.1
q24.3
q25.1
q25.2
q25.3
q26.1
q26.2
q26.3


Pair of chromosomes 15
(light microscope view)
Deletion
15q11.2-13
Chromosome 15
idiogram


NORMAL
ANGELMAN SYNDROME
Genomic imprinting
Maternal copy of UBE3A expressed
Loss of UBE3A expression


Example of Imprinting Inheritance in Familial AS
= UBE3A mutation carrier
= Angelman syndrome
I
II
III
IV
AS
AS
AS

History of Angelman syndrome

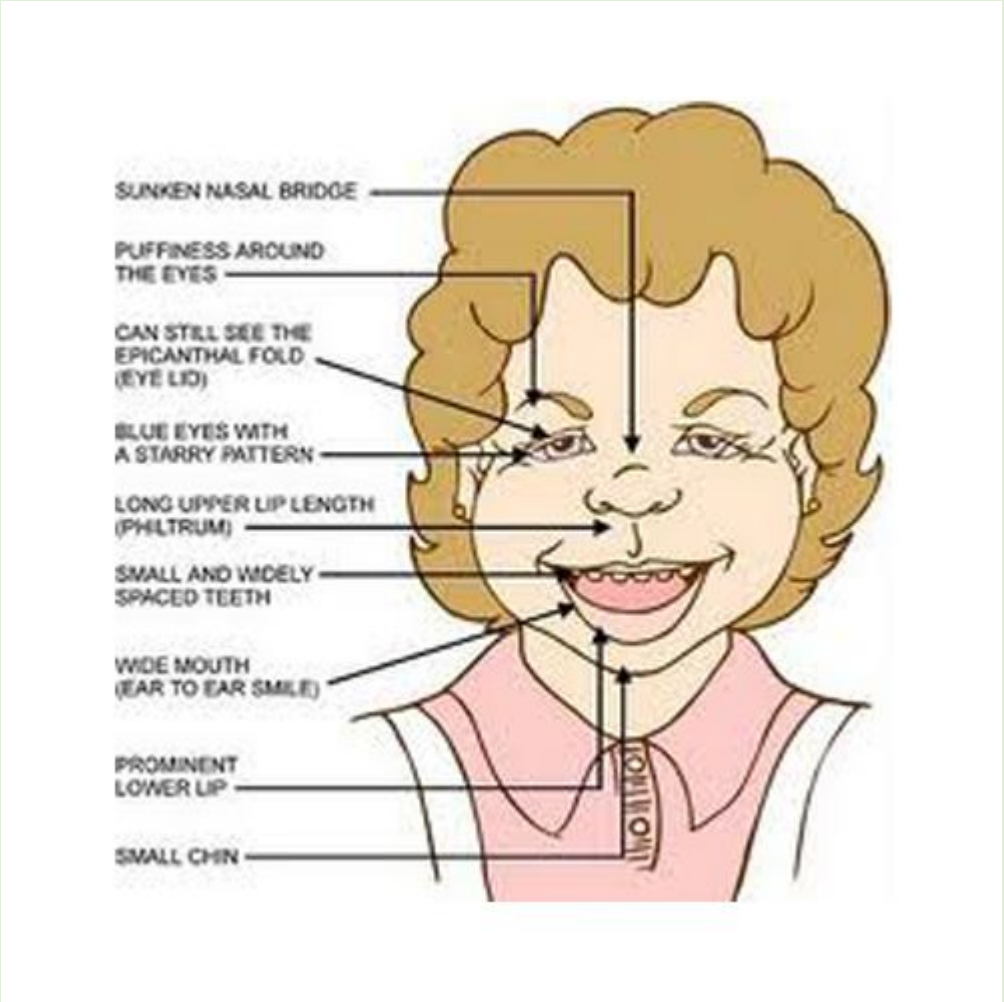
History of Angelman Syndrome

- Angelman Syndrome was diagnosed by Dr. Harry Angelman in 1965.
- Before the diagnosis they called the disorder “Happy Puppet Disorder” because of the way the person moved their arms and hands, and the way they smiled. It looked as if the persons arms were being held up with puppet strings.
- Instead of Angels they were called “Puppet Children”.

GENETICS BEHIND ANGELMAN SYNDROME

Genetics behind Angelman Syndrome

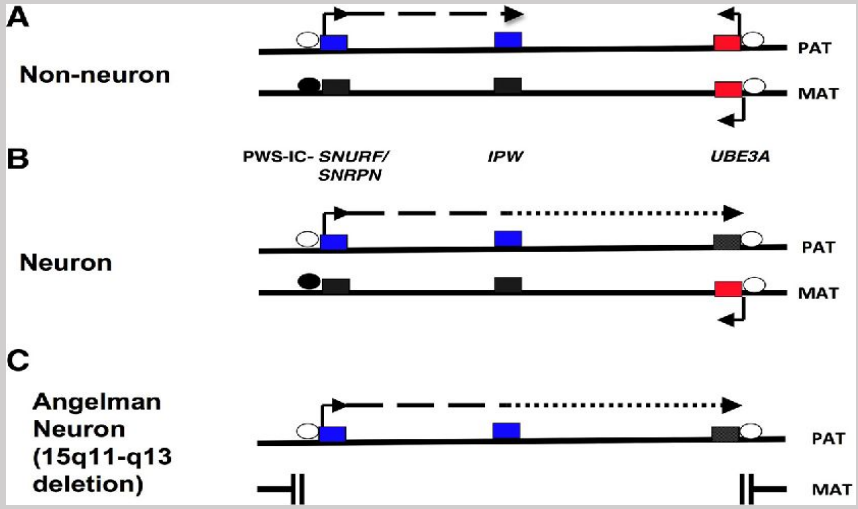
- There are different levels of Angelman Syndrome but no level is hereditary based.
- Most cases of Angelman Syndrome are not inherited.
- It is usually caused by a deletion in the maternal chromosome 15 or by paternal unipaternal disomic (UPD). UPD is when there is 2 copies of the fathers chromosome 15 and the mothers copy is missing.
- In some cases the fathers copies are silenced so you cannot tell at birth that the child has Angelman syndrome (AS)

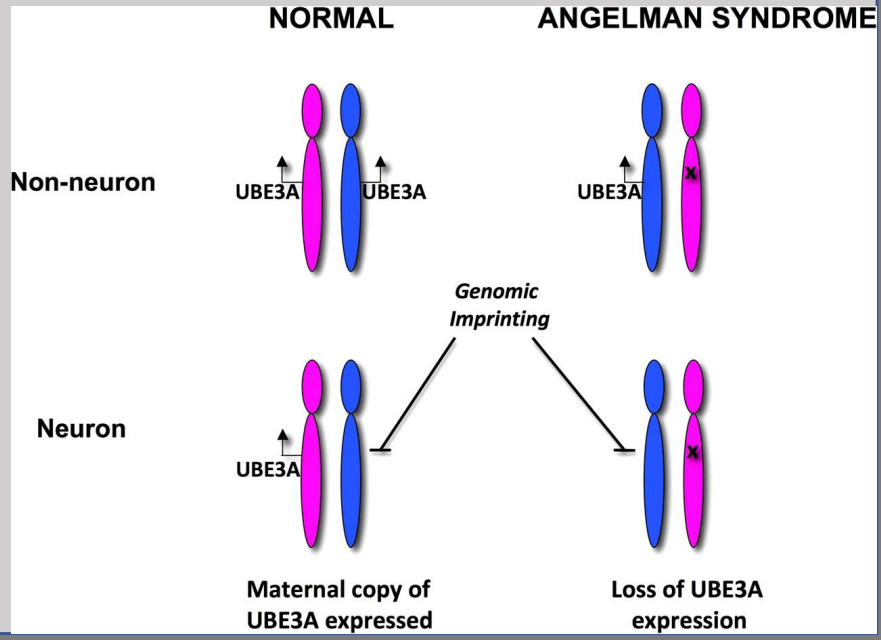


REASONS

Other Causes -

- Angelman Syndrome can also be caused by a chromosomal rearrangement called a translocation, or by a mutation or other defect in the region of DNA that controls activation of the UBE3A gene.
- In these genetic changes it can inactivate the UBE3A or other genes on the maternal side.
- Imprinting: Genomic imprinting refers to a process whereby the maternal copy of a gene can be marked or “imprinted” differently than the paternal copy of the same gene

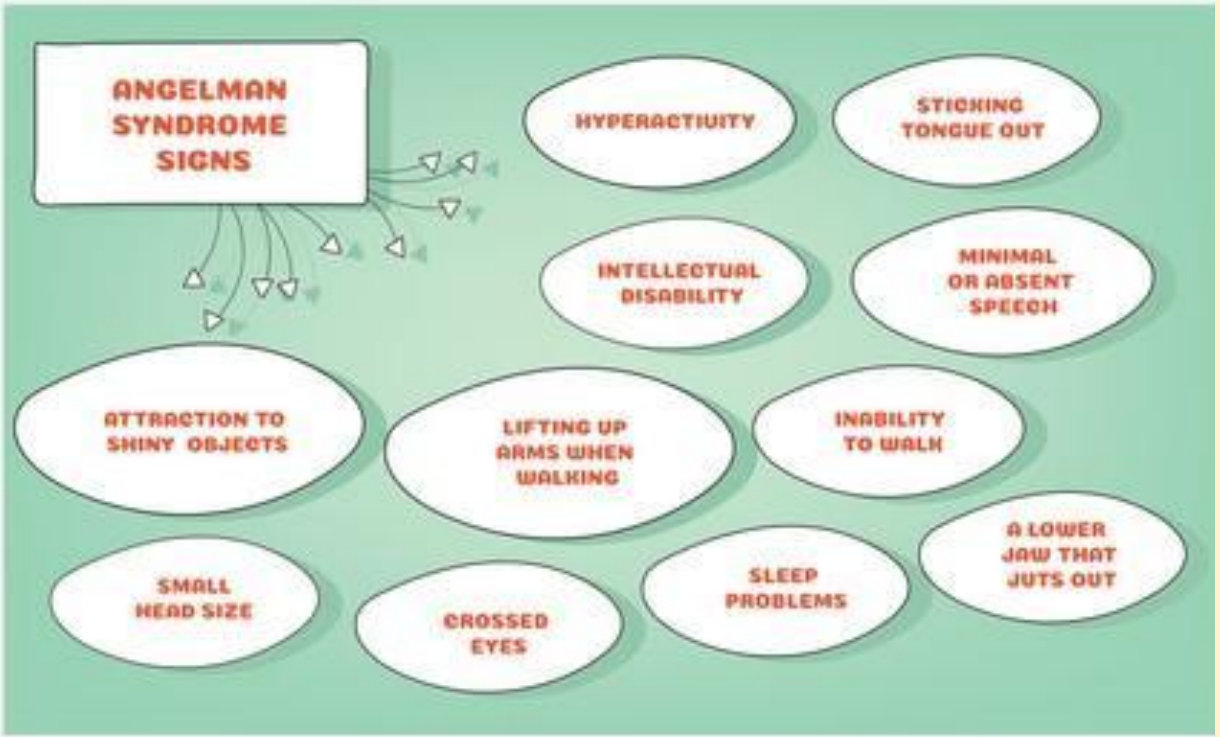




SIGNS OF ANGELMAN SYNDROME

Angelman syndrome signs and symptoms include:

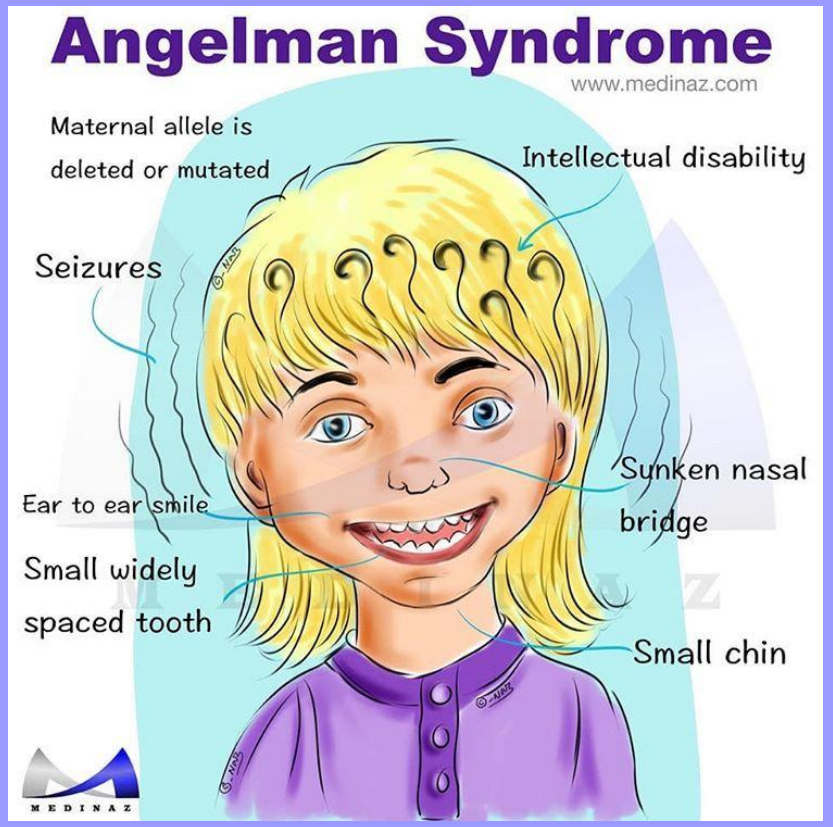
- Developmental delays, including no crawling or babbling at 6 to 12 months
- Intellectual disability
- No speech or minimal speech
- Difficulty walking, moving or balancing well
- Frequent smiling and laughter
- Happy, excitable personality
- Trouble going to sleep and staying asleep



SYMPTOMS

People who have Angelman syndrome may also show the following features:

- Seizures, usually beginning between 2 and 3 years of age
- Stiff or jerky movements
- Small head size, with flatness in the back of the head
- Tongue thrusting
- Hair, skin and eyes that are light in color
- Unusual behaviors, such as hand flapping and arms uplifted while walking
- Sleep problems



OCCURRENCE

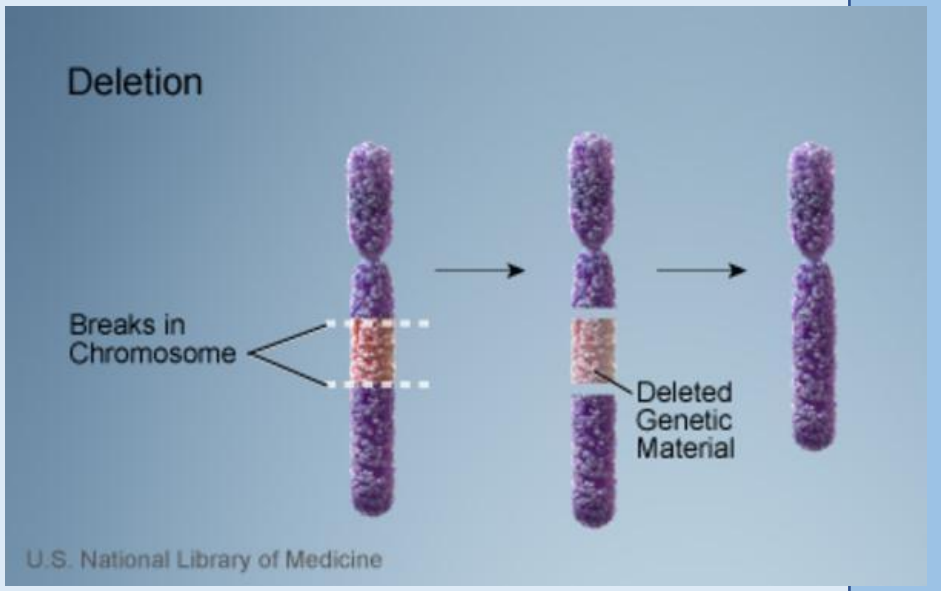
- Babies appear to be normal at birth, but they have noticeable developmental delays when they are 6-12 months old
- Seizures occur between 2-3 years of age
- By the age of 3, kids will have noticeable balance problems, speech impairment, and frequent laughter

CAUSES

Angelman syndrome is a genetic disorder. It's usually caused by problems with a gene located on chromosome 15 called the ubiquitin protein ligase E3A (*UBE3A*) gene.

A missing or defective gene

- You receive your pairs of genes from your parents — one copy from your mother (maternal copy) and the other from your father (paternal copy).
- Your cells typically use information from both copies, but in a small number of genes, only one copy is active.
- Normally, only the maternal copy of the *UBE3A* gene is active in the brain. Most cases of Angelman syndrome occur when part of the maternal copy is missing or damaged
- In a few cases, Angelman syndrome is caused when two paternal copies of the gene are inherited, instead of one from each parent.

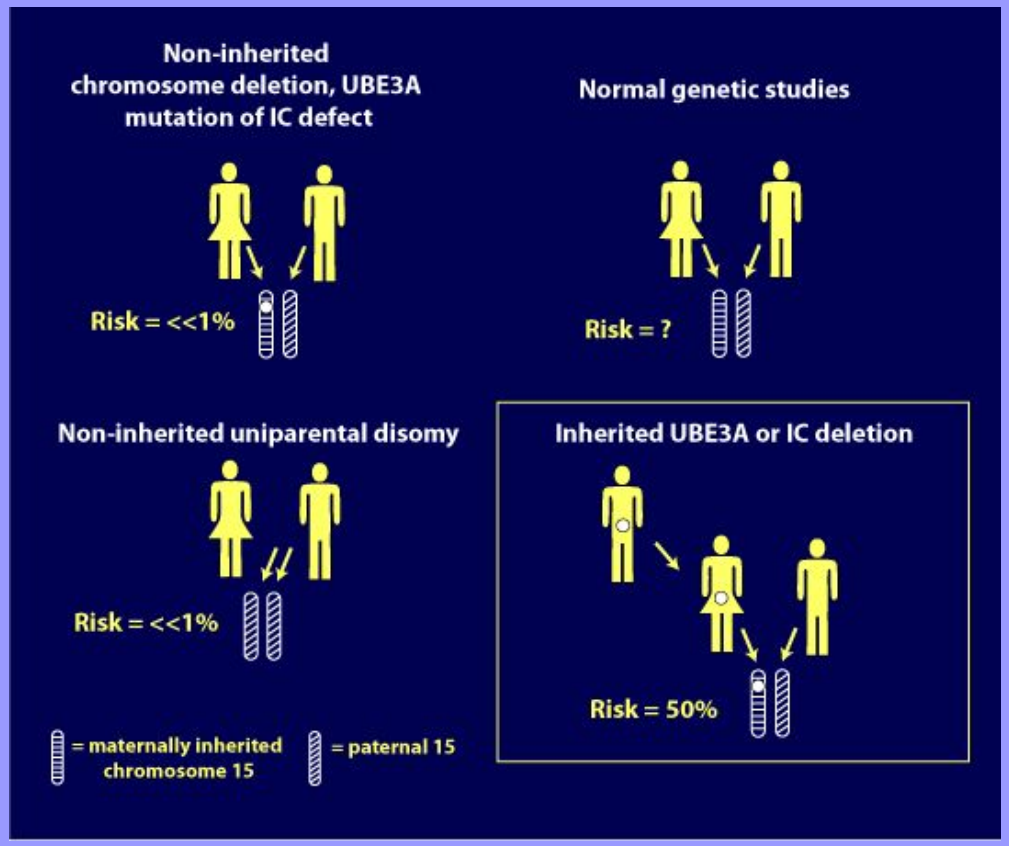


RISK FACTORS

Risk factors

Angelman syndrome is rare. Researchers usually don't know what causes the genetic changes that result in Angelman syndrome. Most people with Angelman syndrome don't have a family history of the disease.

Occasionally, Angelman syndrome may be inherited from a parent. A family history of the disease may increase a baby's risk of developing Angelman syndrome.

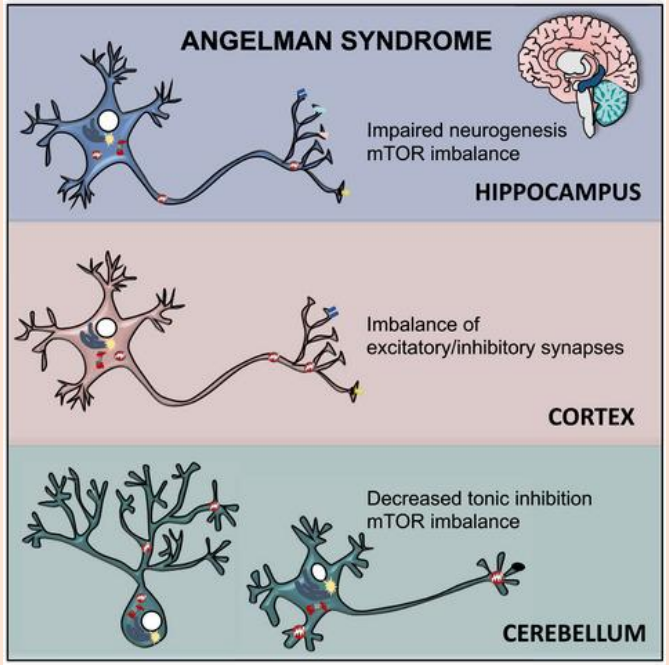


COMPLICATION

Complications

Complications associated with Angelman syndrome include:

- **Feeding difficulties.** Difficulty coordinating sucking and swallowing may cause feeding problems in infants. Your pediatrician may recommend a high-calorie formula to help your baby gain weight.
- **Hyperactivity.** Children with Angelman syndrome often move quickly from one activity to another, have a short attention span, and keep their hands or a toy in their mouths. Hyperactivity often decreases with age, and medication usually isn't necessary.
- **Sleep disorders.** People with Angelman syndrome often have abnormal sleep-wake patterns and may require less sleep than most people. Sleep difficulties may improve with age. Medication and behavior therapy may help control sleep disorders.
- **Curvature of the spine (scoliosis).** Some people with Angelman syndrome develop an abnormal side-to-side spinal curvature over time.
- **Obesity.** Older children with Angelman syndrome tend to have large appetites, which may lead to obesity.



Preventions

In rare cases, Angelman syndrome may be passed from an affected parent to a child through defective genes. If you're concerned about a family history of Angelman syndrome or if you already have a child with the disorder, consider talking to your doctor or a genetic counselor for help planning future pregnancies.

Diagnosis

Your child's doctor may suspect Angelman syndrome if your child has developmental delays and other signs and symptoms of the disorder, such as problems with movement and balance, a small head size, flatness in the back of the head, and frequent laughter.

Tests

A definitive diagnosis can almost always be made through a blood test. This genetic testing can identify abnormalities in your child's chromosomes that indicate Angelman syndrome.

A combination of genetic tests can reveal the chromosome defects related to Angelman syndrome. These tests may review:

- **Parental DNA pattern.** This test, known as a DNA methylation test, screens for three of the four known genetic abnormalities that cause Angelman syndrome.
- **Missing chromosomes.** A chromosomal microarray (CMA) can show if portions of chromosomes are missing.
- **Gene mutation.** Rarely, Angelman syndrome may occur when a person's maternal copy of the *UBE3A* gene is active, but mutated. If results from a DNA methylation test are normal, your child's doctor may order a *UBE3A* gene sequencing test to look for a maternal mutation.

TREATMENT

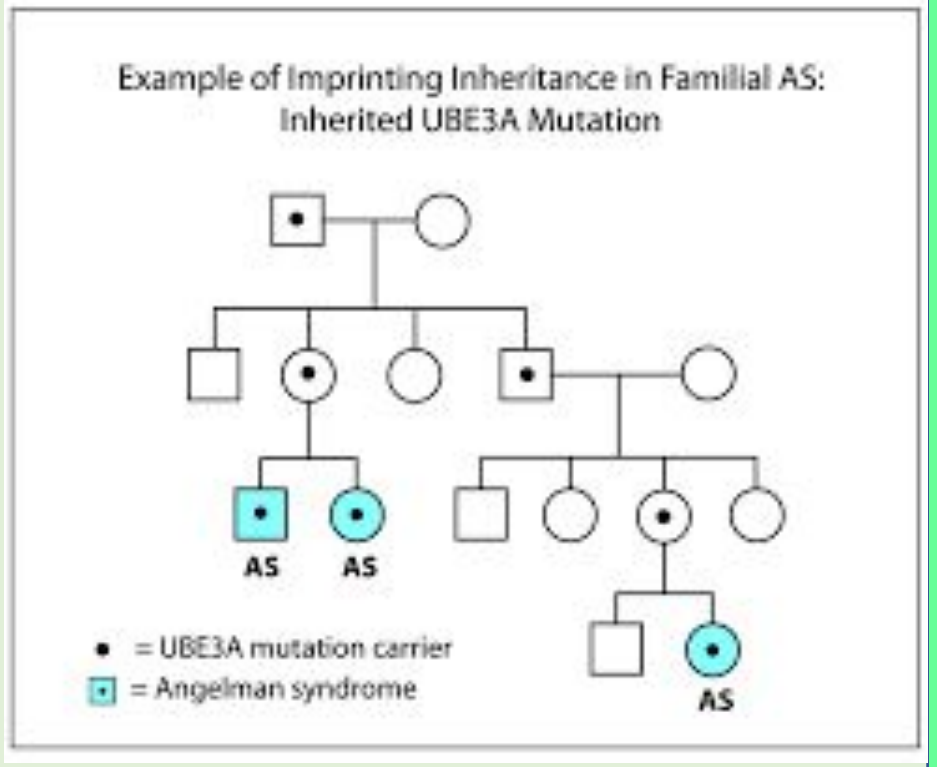
There's no cure for Angelman syndrome. Research is focusing on targeting specific genes for treatment. Current treatment focuses on managing the medical and developmental issues.

A multidisciplinary team of health care professionals will likely work with you to manage your child's condition. Depending on your child's signs and symptoms, treatment for Angelman syndrome may involve:

- **Anti-seizure medication** to control seizures
- **Physical therapy** to help with walking and movement problems
- **Communication therapy,** which may include sign language and picture communication
- **Behavior therapy** to help overcome hyperactivity and a short attention span and to aid in development

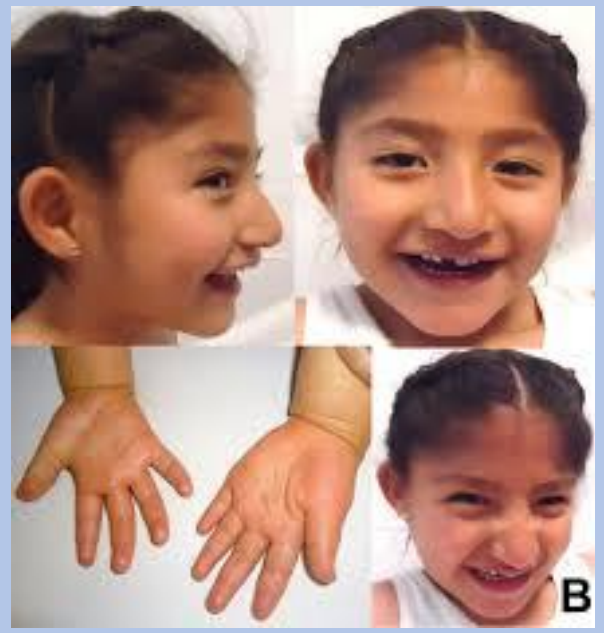
INHERITANCE

- Most cases of Angelman syndrome are not inherited, particularly those caused by a deletion in the maternal chromosome 15 or by paternal uniparental disomy .
- These genetic changes occur as random events during the formation of reproductive cells (eggs and sperm) or in early embryonic development. Affected people typically have no history of the disorder in their family.
- Rarely, a genetic change responsible for Angelman syndrome can be inherited. For example, it is possible for a mutation in the *UBE3A* gene or in the nearby region of DNA that controls gene activation to be passed from one generation to the next.



LIFE EXPECTANCY

Some symptoms of Angelman syndrome become more severe as individuals get older. Sleep issues and seizures tend to become less severe or infrequent. Because of mobility issues, obesity and scoliosis can develop in adolescence.
The life expectancy of people with Angelman syndrome is normal. Angelman syndrome itself does not cause death. However, there can be severe complications due to some of the symptoms of the syndrome, such as seizures and aspiration pneumonia. There is also the possibility of accidents due to walking and balance issues and attraction to water that can cause severe injury.
Individuals with AS will require life-long care, but can live long, happy lives.



A day when all organizations and families gather together and raise awareness about Angelman Syndrome.

INTERNATIONAL ANGELMAN DAY FEB 15

Angelman Today

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www.angelmansyndrome.org
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INTERNATIONAL ANGELMAN DAY FEB 15

WORKSHEET

Nidhi – what is Angelman Syndrome
which chromosome does it affect?
Sakhi – What kind of defect arises in the chromosome to cause Angelman Syndrome?
Angelman Syndrome is also known as?
Keerthana - Who diagnosed Angelman Syndrome?
why is it called Happy Puppet disorder?
Ekta – instead of angels what is the other name given to people with Angelman Syndrome?
What do you understand by UPD?
Teena- Explain the genetics behind Angelman syndrome?
when do we celebrate International Angelman day?
Amit – Explain the signs in Angelman Syndrome?
at what age the occurrence of Angelman syndrome is visible?

Karmshil - What are the causes behind Angelman Syndrome?
Is Angelman Syndrome, inherited?
Harish- Explain the symptoms of Angelman Syndrome ?
Draw a pedigree chart demonstrating Angelman Syndrome?
Hari Shankar- What are the risk factors in Angelman Syndrome?
What are the preventive measures to follow if someone has Angelman Syndrome?
Aishwary - What are the complications in Angelman Syndrome?
write about relation of Angelman Syndrome in inheritance?
Ashwin- How can we diagnose Angelman Syndrome?
write about the treatment given to someone with
, Angelman Syndrome?
Vikram- what is the life expectancy of someone with Angelman Syndrome?
Is their any cure to Angelman Syndrome?

Thank you For watching

I hope you learned something today