

Angel man Syndrome

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Abstract

Angel man syndrome is a rare genetic and neurological disorder which is characterized by severe developmental delays and learning disabilities. Although, those diagnosed with the syndrome may be unable to speak, but many gradually learn to communicate through other means such as sign language. Additional symptoms also include seizures, sleep disorders and feeding difficulties. Some children with Angel man syndrome may have distinctive facial features, but mainly facial features reflect the normal parental traits. Angel man syndrome is caused by deletion or abnormal expression of the UBE3A gene.

Keywords: Angel man syndrome, UBE3A gene, seizures.

1. Introduction

Children are an asset for future National abundance. Talking statically, there are 73.8 million children in the world of aged 0-17 in years. As children under five are more vulnerable to various infections, they are at the risk of high mortality and morbidity rates all over the world. Considering, them as an assets to nation, it is the responsibility of the particular government to provide comprehensive health services to all the Children to promote the health and wellness.

Angel man syndrome was first described in medical literature in 1965 by Dr. Harry Angel man, an English Physician which was initially termed it as “**Happy Puppet Syndrome**” characterized with cheerful facial expression and jerky movements of the afflicted children, and the diagnosis of the disorder is usually made between 1 to 4 years of age.

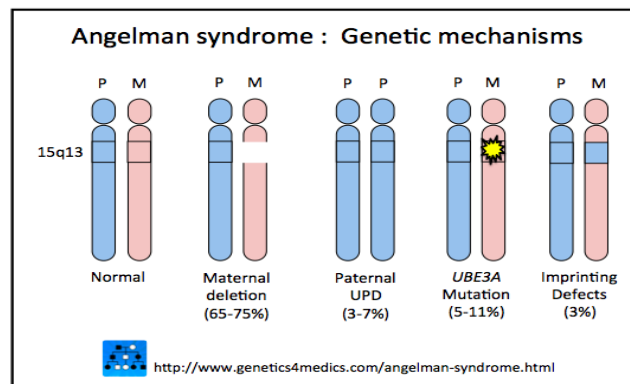
1.1 Definition:-

Angel man Syndrome (AS) is a genetic disorder that mainly affects the nervous system, characterized by symptoms such as; they are usually happy and have a particular interest in water. The symptoms are usually noticeable by age of one.

1.2 Incidence -

Angel man syndrome affects 1 in 12,000 to 20,000 population, both males and females are equally frequently affected.

1.3 Causes:-



Angel man 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.

- Problems are associated with location of gene on Chromosome 15 called ubiquitin protein ligase E3A (UBE3A) gene.
- A missing or defective gene copy of genesis usually missing especially the maternal copy.
- In rare cases, inheritance of two paternal copy of gene from the same parent, instead of one from each parent.

1.1 Risk factors:-

- According to researchers, it is often idiopathic.
- Angel man syndrome is rare.
- In many cases, family histories are not found among angel man syndrome child.

1.5 Signs and Symptoms:-

Consistent Signs and symptoms :- (100%)

- ✓ Developmental delay, functionally severe
- ✓ Speech impairment :- No or minimal use of words, receptive and non-verbal communication skills higher than verbal communication
- ✓ Movement or Balance disorder: - Ataxia of gait, and tremulous movement of limbs.
- ✓ Behavioral Uniqueness :- Combination of frequent laughter or smiling, happy appearance, easily excitable personality, often hand flapping movements, hyper motoric behavior, short attention span.

Frequent signs and symptoms :- (more than 80%)

- ✓ Delayed, disproportionate growth in head circumference results in microcephaly by age 2.
- ✓ Seizures usually at age of 3.
- ✓ Abnormal EEG

Associated signs and symptoms :- (20-80%)

- ✓ Strabismus :- It is a disorder in which the eyes don't look in exactly the same direction at the same time
- ✓ Hypo pigmented eyes and skin
- ✓ Tongue thrusting: - Tongue protrudes through the anterior incisors during swallowing, speech and while tongue is at rest which cause suck and swallowing disorder.
- ✓ Hyper active tendon reflexes
- ✓ Feeding problems during pregnancy
- ✓ Uplifted, flexed arms during walking
- ✓ Increased sensitivity to heat

1.6 Diagnosis:-

The diagnosis of Angel man syndrome is based on:

- Brief health assessment including developmental assessment especially in area of verbal development.
- Recording of unusual movements including fine tremors, jerky limb movements, hand flapping

- A history of epilepsy and abnormal EEG findings.
- A happy appearance with frequent laughter
- Gene mapping will reveal - Parenteral DNA Pattern and to identify missing chromosome to find out gene mutation.

1.7 Treatment:-

Treatment strategies include -

- **Anti seizure therapy** – helps to control seizures
- **Physical therapy** - help with walking problems.
- **Communication therapy** - which may include sign language and picture communication.
- **Behaviour therapy** – helps to overcome hyper activity and short attention span.

1.8 Complications:-

- **Feeding difficulties :-**
- ✓ Difficulty in coordination of sucking and swallowing.
- ✓ High calorie formula feeds to maintain weight of baby.
- **Hyperactivity :-**
- ✓ Can be maintained by play therapy
- ✓ It often decreases with age and doesn't require any medicine intervention.
- **Sleep disorders :-**
- ✓ Abnormal sleep-awake patterns improves with age
- ✓ Medication and behavior therapy may be effective.
- **Scoliosis**
- **Obesity**

2. Role of a nurse in managing child with angel man syndrome and their family -

- ❖ Nurse play a key role in dealing the patients with angel man syndrome and even can help the affected families to deal with the changes in behavior and developmental milestones of the affected child.
- ❖ Nurse should assist the child to develop their potentialities in self care management by appropriate guidance and supervision
- ❖ Nurse should alleviate the anxiety of the parents of affected children by helping them to cope up with the situation by providing support groups and counseling Services.
- ❖ Nurse should help the families to collaborate with the health care team for child proper development and treatment.
- ❖ Nurse should counsel the parents for genetic test while planning second child if they had already a child with angel man syndrome.

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