Angelman Syndrome

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NARRATIVE REVIEW

Abstract

Angelman syndrome (AS) is a rare disorder characterized by ataxia, ambulation difficulties, muteness, excessive laughter and seizures. Most children diagnosed with (AS) have an abnormal head computed tomography scan, an abnormal electroencephalogram, and an abnormal pneumoencephalogram. They exhibit a range of dental disorders that directly affects their oral health related quality of life. This syndrome profile details the salient features of AS.

Keywords: *Angleman Syndrome, Imprinting Disorder, Puppet Children, Happy Puppet Syndrome, Ataxia, Chromosome Disorders*

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History

Angelman Syndrome is a neurogenetic disorder that was first described by Harry Angelman in 1965. It was initially known as 'happy puppet syndrome' as Angelman first described his subjects as 'puppet children'. Angelman syndrome's latest prevalence of 1:15000 was recorded in 1995.

Etiology

It is caused by lack of expression of maternal ubiquitin-protein ligase E3A (UBE3A) gene in the brain. This can be due to the following reasons:

- Deletion in chromosome 15q11-q13 (Figure 1)
- Point Mutation
- Imprinting Defect

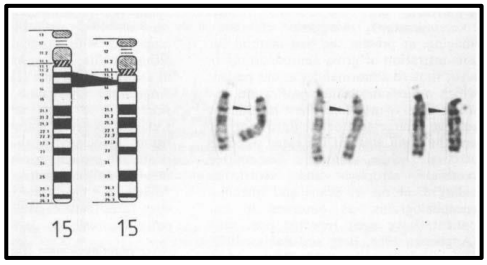

Figure 1: Yamada KA and Volpe JJ. Developmental Medicine & Child Neurology. 1990;32(11)

- Paternal uniparental disomy

Characteristics

- Intellectual disability (IQ <25)
- Speech disorders
- Language delay
- Cognition deficits
- Sleep difficulties
- Unusually happy demeanour
- Paroxysmal laughter
- Seizure activity
- Delayed onset of sitting (12 months) and walking (3 year)

Orofacial Features

Facial: Blue eyes, Fair/blond hair, Microcephaly, Elongated face (Figure 2), Disproportionate facial thirds, Flat occiput, Deep-set eyes, Strabismus

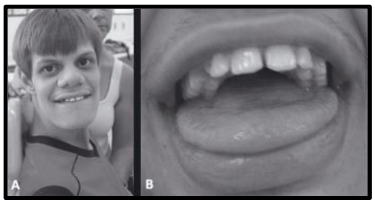

Figure 2 Source: de Queiroz AM et al. Special Care in Dentistry. 2012:33(1)

Dental: Maxillary hypoplasia, Prognathism, Protruding Tongue, Tongue thrusting, Sucking and Swallowing disorders, Excessive chewing and drooling, Feeding difficulties during infancy, Hypomineralization of primary molars, Single-root molar, Mouth breathing

Management

Only supportive treatment exists, with the main focus being mitigation of symptoms that affect quality of life. During a dental treatment, physical restrain and mouth prop maybe needed to control involuntary limb movement. In worse cases, treatment under general anesthesia is recommended.

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