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CASE REPORT

ANGELMAN SYNDROME: A CASE REPORT

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ABSTRACT

Angelman syndrome (AS) is a neurodevelopmental disorder characterised by severe learning difficulties,ataxia, a seizure disorder with a characteristic EEG, subtle dysmorphic facial features, and a happy,sociable disposition and a behavioral uniqueness. Children with AS shows oral manifestations like tongue thrusting, diastema, mandibular prognathism, frequent drooling and excessive chewing behavior.This case report elaborates a child patient of 5 yrs old suffering from AS who reported to the clinics with a chief complain of pain and decayed teeth. The child was treated comprehensively with pulp therapy, Stainless steel crown, extraction and apt space management.

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INTRODUCTION

WHO defines a handicapped individual as one who, over an appreciable time is prevented by a physical or mental condition from full participation in the normal activities of his age group, including those of a social, recreational education and vocational nature. Angelman syndrome is a genetic disorder characterized by severe developmental delay, speech disorder, ataxia, craniofacial abnormalities, and odd behavior accompanied by a happy disposition and occasional bouts of uncontrollable laughter (Williams, 2006 and Valente, 2006). An English pediatrician Harry Angelman, in 1965, reported clinical findings in three children with similar features of severe learning disability, ataxic, jerky movements, inability to speak, and easily provoked laughter. All three had epileptic seizures with a characteristic EEG appearance and subtle dysmorphic facial features (Angelman, 1965). This condition was originally known as the "happy puppet" syndrome and is now known by the less derisive term of Angelman syndrome (Landsman, 2012). There is no reported gender predominance. The incidence of the disease varies from one in 10,000 to one in 40,000, with speculation that the disease is underdiagnosed due to its various phenotypes (Buckley, 1998). For many years, it was considered a rare disorder, although the occurrence of families with affected siblings suggested a genetic etiology, no known cause could initially be identified. It is usually diagnosed around two years of age, but because they developed milder clinical manifestations of the disease, there are instances of patients not being diagnosed until their early teens (Ramanathan, 2008).

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Pathogenesis

There are 4 known genetic mechanisms that result in Angelman syndrome (Williams, 2006; Maguire, 2009 and Varela, 2004)

- Molecular deletions involving the meiotically unstable 15q11.2-q13 critical region (65% - 75% of cases)
- Paternal uniparental disomy (UPD)

- Imprinting defects (IDs)
- Mutations in the ubiquitin-protein ligase E3A gene (mUBE3A).

Clinical Features

Individuals with AS usually present with the following clinical features

- Developmental delay;
- Speech impairment with minimal or no use of words(receptive and nonverbal communication skills are higher than verbal ones);
- Unique Behavior - Frequent laughter/smiling, apparent happy demeanor, easily excitable personality, hyperactivity and short attention span;
- Head circumference growth is delayed; usually resulting in microcephaly by age of 2 years;
- Seizures, onset usually before 3 years of age; and
- A characteristic EEG patternwhich is abnormal with large amplitude, slow spike waves.

Associated clinical features of children with AS like tongue thrusting, mandibular prognathism, diastemas, a thin upper lip, (Brown, 2004) micro- or brachycephaly, (http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=10583) and excessive chewing behaviorare of extreme importance to dentists and are characteristics that the child presents, which may require direct or indirect intervention, with treatment such as orthodontics. Other characteristics like hyperactive tendon reflexes, frequent drooling, sucking/ swallowing disorders, increased sensitivity to heat, sleep disturbances, and fascination with water and reflective surfaces (Clayton-Smith, 2003) may be helpful or disruptive during dental treatment. As a health professional, the dentist must also be aware that hypopigmented skin and eyes,strabismus, uplifted and flexed arms during walking and a flat back of the head may be present in patients with AS. As with any other child, it is essential to consider the psychological aspects of the dental treatment of children with AS in order to achieve successful behavior management and, consequently, successful dental treatment. Developmental delay, speech impairment, hyperactivity, and a short attention span are characteristics of children with AS, which make behavior management extremely challenging. Yet, as with the clinical case described in this paper, with the right techniques and psychological approach, it is possible to treat children with AS from a very young age without the need for general anesthesia.

Case Report

A 5 year-old female patient with AS presentedfor dental treatment to the Department of Pediatric and Preventive Dentistry, M. A. Rangoonwala Dental College and Research Centre, Pune. There was no consanguineous relationship between the parents.She was born at term gestation through LSCS after an antenatal period complicated by maternal hypothyroidism and hypertension. The diagnosis of AS was confirmed by clinical and laboratory criteria, in which a mutation of the maternal chromosome 15q11-13 was found using FISH test. No contributory prenatal history was found, during the clinical interview.According to the parents, the patient experienced a delay in learning motor skills. Currently, the patient showed severe intellectual disability, absence of speech, a severe inability to maintain attention, frequent laughter, and an easily excitable personality with an affectionate nature. In addition, the patient exhibited unintentional movements of his arms and hands. Craniofacial examination revealed microbrachycephaly(head circumference 3^{rd}-5^{th} centile), normal hair, hypopigmented skin, a large mouth with a thin upper lip, disproportion of the facial thirds, deeply set eyes, tongue protrusion and Strabismus (Figure 1).

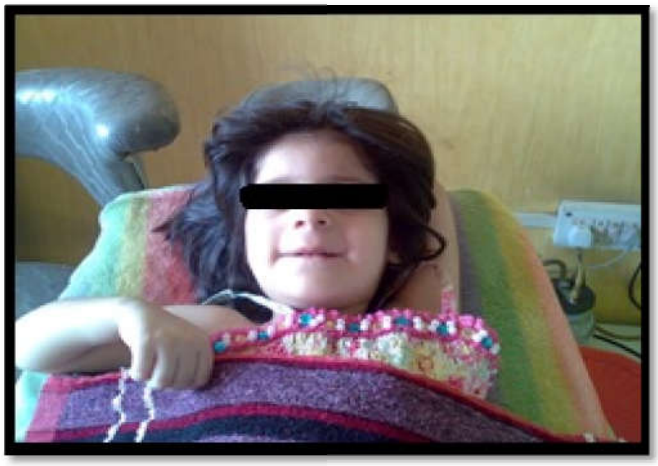

Figure 1. Extraoral Frontal View

Intraroral clinical examination revealed poor oral hygiene (OHI-S) (Greene, 1964) and extensive carious lesions, with cavitation involving the posterior teeth (Figure 2 and 3).

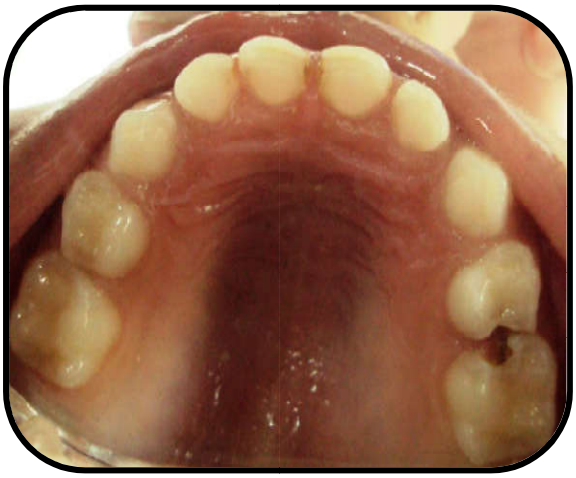

Figure 2. Preoperative Maxillary Arch

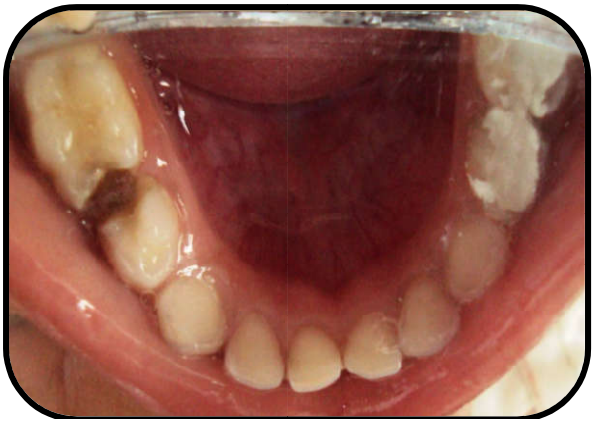

Figure 3. Preoperative Mandibular Arch

The patient was uncooperative and reluctant to receive treatment (category 2 of Frankl's behavior rating scale). Behavior management techniques were employed as recommended by the AAPD's Guideline on Behavior Guidance for the Pediatric Dental Patient (American Academy of Pediatric Dentistry, 2011), (communication and communicative guidance, tell-show-do, voice control, nonverbal communication, positive reinforcement, distraction, and desensitization). Mouth props and bite blocks were used for all oral procedures to keep the mouth opened. A consent form was signed by the parents of the child before implementation of all procedures. The combination of behavior guidance, mouth props and proper isolation allowed dental care to be accomplished under moisture-controlled conditions and in an effective and safe manner for the patient and dental team. The treatment plan included oral hygiene instructions and diet counselling, followed by oral prophylaxis. Four periapical radiographs, one of each posterior region, were taken with the aid of a film holder. Pulp Therapy was performed with 4 teeth (55, 74, 75, 85) (Figure 4 and 5) followed by restoration with Glass Ionomer Cement and Stainless Steel Crown (3M).

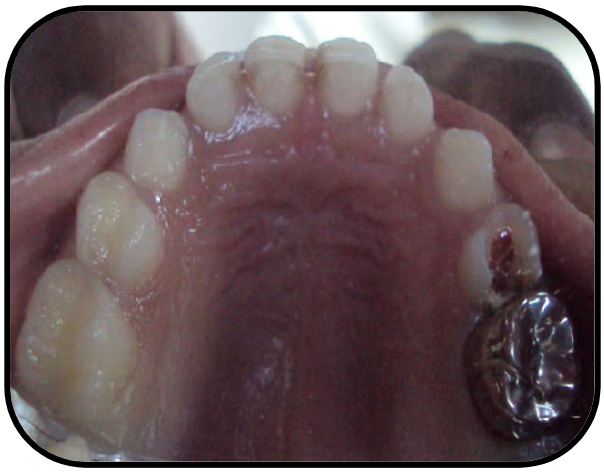

Figure 4. Post Operative Maxillary Arch

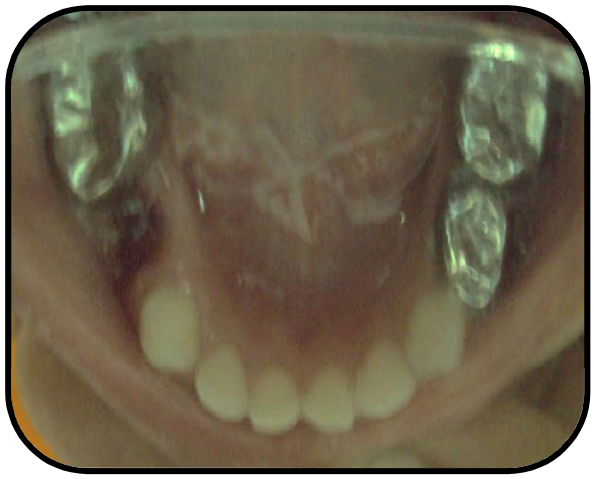

Figure 5. Post operative Mandibular Arch

One primary molar (84) was extracted as the tooth was severely destroyed by caries, followed by a crown and loop space maintainer (Figure 6). All restorative procedures were performed under infiltration/block of one carpule of local anesthesia and with proper isolation technique. The patient returned to the dental clinic every 4 months for a period of 2 years to receive prophylaxis. After eruption of first permanent molars, topical fluoride application and sealant placement was done as primary prevention. After that period, the patient returned every year for clinical and radiographic follow-ups.

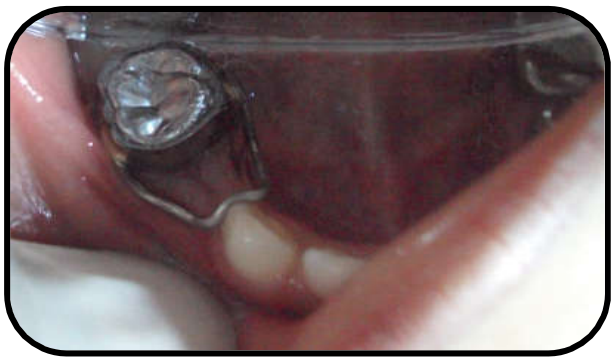

Figure 6. Band and Loop Space Maintainer

DISCUSSION

As with any other child, the psychological aspects of dental treatment in children with AS should be addressed to achieve successful behavior management and a favorable treatment outcome. [15]Children with AS should return to the dentist every 4 months for re-evaluation, preventive treatment and to maintain their familiarity with the dentist.If good communication is established between the parents, the dentist and the child, then cooperation during the treatment is possible. Children with AS who are receiving medical treatment with drugs, should have their dental visits scheduled in the early morning to benefit from the maximum effect of the medication. Simple and clear instruction should be given to the child and should be repeated many times. While instructing the patient eye contact should be maintained to prevent the child from getting distracted. Knowing that children with AS are fascinated with water (http://www.ncbi.nlm.nih.gov/ entrez/dispomim.cgi?id=10583), crinkly items, (Williams, 2006; http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi? id=105830) and reflective surfaces (Clayton-Smith, 2003) the dentist may use water and mirrors when trying to familiarize the child with the dental environment and procedures in order to manage the child's behavior. Intensive Oral Hygiene Education must be given both to the child and to the caretaker, as the child has involuntary movements and lacks the motor coordination necessary to brush correctly independently. Special emphasis should be given to the brushing technique. Since multicolored stimuli make the child excited and distracted, colorful and bright objects (Red and Yellow) should be avoided in the dental office. Constant and frequent compliments are helpful. If the parents are not motivated to become conscientious and compliant, and they do not maintain good home care, it is very difficult to maintain the child caries-free in spite of frequent dental visits. With regular visits and with the cooperation and dedication of the caretakers, it is possible for a child with AS to remain caries-free.

Conclusion

Angelman syndrome affected children show multiple dental manifestations as a result of the syndrome itself and also due to movement abnormalities like ataxia and involuntary movement results in inability to maintain oral hygiene. They require behavioral modification and guidance to both the child and the parent to make the child cooperative for dental treatment

which should be acceptable to the parent and of highest quality.Patient education, motivation and establishing a positive relationship with the child and his/her family should be the ideal motive of every pediatric dentist alone with the establishment of Dental Home.

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