Angelman syndrome : A rare case report

ABSTRACT:

Angelman syndrome (AS) is a rare neurodevelopmental disorder characterized by severe intellectual disability, speech delay, dysmorphic features, cognitive impairment, seizures as well as a unique behaviour with an inappropriate happy demeanor. This article discuss the case of an eight year old male patient with the chief complain of dental pain but presented with the signs of AS. He appeared hyperactive and easily excitable with an unusual laughing facial expression. His speech was impaired and showed unusual fascination towards water. Metabolic screening tests and brain Magnetic Resonance Imaging (MRI) revealed no abnormality. Genetic analysis is pathognomonic for Angelman Syndrome.

Key words: Angelman Syndrome; Neurogenetic Disorder; Myoclonus; Epilepsy

Introduction:

Angelman Syndrome (AS) was first described in 1965 by English paediatrician Dr. Harry Angelman as "happy puppet" syndrome.[1] It is a neurogenetic disorder caused due to loss of expression of maternal UBE3A protein, encoded on chromosome 15q11.2–13.1, which result in distinctive phenotype.[2]

Development delays are usually evident within the first year of life. The behavioural features include easily provoked laughter, short attention span, hypermotoric behaviour, mouthing of objects, sleep disturbance and affinity towards water. Movement disorders include jerkiness, ataxic gait and tremors.[3] The incidence of AS is 1:10,000 to 40,000 with no gender predilection. [4-5] This case describe a child with AS.

Case Report:

An eight year old male patient reported with complaint of pain in maxillary left posterior region since 3 days. Pain was spontaneous in origin, dull and continuous in nature. It aggravated on its own and relieved by taking medication.

Child was hyperactive and presented with unique behaviour of constant smiling, frequent laughing and excitability. He showed fascination to water/crinkly items. He could walk on his own but with shrugged shoulders and slightly raised arms flexed at the elbows (Figure 1).

Figure 1: Profile picture of a child with Angelman syndrome.

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He was 112 cm tall and 23.2 kg in weight. He was the third child of nonconsanguineous healthy parents. Also, there was no history of allergy and seizures but IQ level was below than normal. He was born at the time via vaginal delivery with normal Appearance, Pulse, Grimace, Activity, and Respiration (APGAR) score. He had a happy face and the parents noticed his hyperactivity at the age of 5 years. They reported that he was able to sit at the age of 7 months and was able to walk independently at the age of 14 months. They also described his disturbed sleep-wake cycle wherein the patient sleeps late at night and wakes up early morning.

On physical examination, the child presented with dolicocephalic head, flat occiput, mandibular prognathism, clubbing in fingers of both the hands, drooling of saliva and difficulty in articulated speech. Intraorally, patient revealed partially bifid tongue, multiple carious teeth and mammelons in incisors (Figure 2 and 3). Overall the child was hyperactive with impaired speech, sleep disturbance, excitability and fascination with water. His muscle tone, force and deep tendon reflexes were normal. Besides, he could walk and run independently.

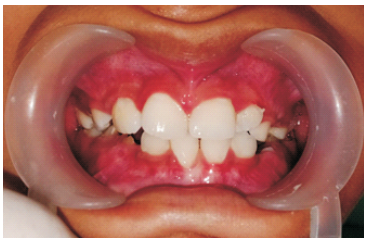

Figure 2: Intraoral image

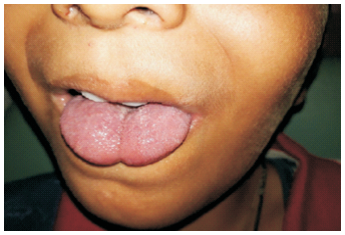

Figure 3: Image reveal bifid tongue

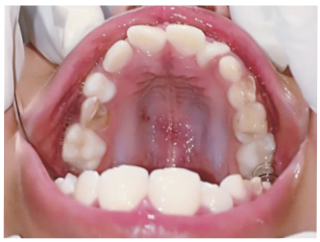

Figure 4: Pre operative intraoral maxillary view

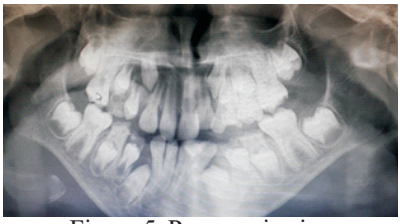

Figure 5: Panoramic view

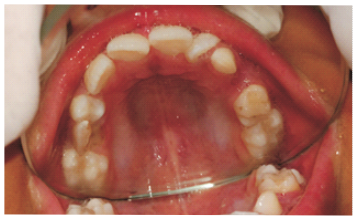

Figure 6: Post operative view reveals extraction irt. 64

Panoramic radiograph revealed multiple retained teeth, unerupted permanent teeth and reduced height of alveolar bone (Figure 5). Laboratory findings were normal. Electroencephalography (EEG), Electrocardiography (ECG), Chest radiograph and long bones radiographs appeared normal.

Treatment of the patient was planned out and approached systematically starting with oral prophylaxis along with continuous monitoring of periodontal health. This was followed by the extraction of deep carious teeth 64, 74, 75, 84, 85 (Figure 6). The patient was later subjected to orthodontic treatment for aesthetic correction.

Table–1: Clinical Features of Angelman Syndrome [3].

Consistent	Frequent	Occasional
Functionally severe intellectual disability	Microcephaly with flat occiput/occipital groove	Scoliosis
Movement/balance disorder	Seizures	Hypopigmentation
Speech impairment	Abnormal EEG	Increased sensitivity to heat
Behavioral phenotype (easily excited, happy, frequent laughter, hypermotoric)	Gastrointestinal difficulties (feeding problems, gastroesophageal reflux, constipation)	Growth disturbance depending on genotype
	Fascination with water or crinkly items	Ocular problems (refractive and alignment errors)
	Mouthing behavior	
	Ankle pronation	
	Sleep disturbance	

Discussion:

AS can be diagnosed from 2 years of age up till adulthood due to lack of overtly distinctive phenotype presentations.[6] The peculiar behavioral profile mostly become evident in second and third years of life, that is when the most common dysmorphic features are observed.[7] In this case, parents of the patient identified the developmental delay when the patient was 3 years of age.

These patients are commonly referred as having "happy puppet syndrome" because of their happy disposition, frequent and inappropriate bursts of laughter along with the hand-flapping and trembling gait.[6]

Excessive salivation can be due to lack of swallowing which can cause drooling. It can also be caused by stomatitis or use of certain medications.[8] The child with AS presented high risk of caries incidence because of inability to maintain proper hygiene due to lack of motor coordination and cognitive impairment; regular use of medications that can be sugared and cause xerostomy; an uncontrolled and cariogenic diet; and difficulty in accessing regular professional dental care.[9] The patient who have AS shows craniofacial abnormalities (Table 1) including microcephaly, high-arched palate, tongue protrusion with thrusting, macroglossia, diastemas, excessive chewing behavior, wide jaw and flat occiput, all of which contribute to potential airway difficulties in this patient population. However, even though our patient presented with few such abnormalities (Table 2) like wide mouth, abnormal teeth, tongue protrusion causing bifid tongue and mandibular prognathism, he fortunately did not experience any airway complications.

Impaired communication is mainly due to the symptoms of language deficit and mental retardation. Even our patient seemed mentally retarded with speech defect. Additionally, clinical features of AS include hyperactive tendon reflexes, sleep disturbances, short attention span, strabismus, obesity, sucking/swallowing disorders and history of epilepsy.

Gastrointestinal distress is one of the most critical issue that require clinical care for many individuals across all age groups in AS.[2] The medications for the same (if under any) may add in complexity to the anaesthesia management of the patient. Usually, the child with AS shows appearance of striking high voltage slow-wave activity is a frequent characteristic finding on EEG.[10] In our case report though, EEG and ECG findings were normal and gratefully no such complications arose.

Conclusion :

Angelman syndrome affected children present with myriad challenges to a clinician. Apart from behavioural and medical consideration, oral intervention is also requisite. Multiple oral manifestations are presented in patients with Angelman Syndrome as a consequence to not just the syndrome but also the movement abnormalities like ataxia and myocopus which results in inability in maintaining proper oral hygiene. Behavioural modification and guidance is required for both the child and the parent to proceed with any dental procedure which is 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 to the clinician along with the establishment of Dental Home

Table 2: Oral manifestations of Angelman syndrome found in the dental literature[8]

Author	Year	Cases (n)	Age (years)	Oral manifestations
Murakami, et al	2008	1	From 4 to 15	Habit of pacifier sucking, open bite, prominent diastema between central incisors and eruption delays.
Ramanathan, et al	2008	1	5	High arched palate, poor oral motor control, difficulty controlling his saliva, blowing / sucking and with mouth closure.
Sarkar, et al	2011	1	6	Caries in upper incisors and in temporary molars, heavy plaque and stain deposits indicate poor oral hygiene.
Gallo, et al	2012	8	Average 6.8	Wide mouth and jaws, diastemas, insertion of the tip of the tongue between front teeth and front open bite, frequent drooling, sucking, eating and drinking difficulties, enamel erosions, incipient tooth lesions, disturbancies in tooth eruption, mandibular prognathism with anterior cross bite and deep dental caries on primary molars.
de Queiroz, et al	2013	3	Average 12	Prominent mandible (prognathia), large mouth, widely spaced teeth, thin upper lip, tongue protrusion, excessive chewing behavior, excessive drooling, enhanced cheeks, mouth breathing, long and narrow face, open mouth, lowered mandible position, disproportion of facial thirds and hypotonia of tongue and lips.

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