Tufts University School of Dental Medicine

Smith-Magenis Syndrome: A Case Report

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Abstract

Smith-Magenis syndrome is a developmental disorder that affects many parts of the body. The condition results from the deletion of a small piece of chromosome 17 p11.2 resulting in haploinsufficiency for the gene retinoic acid-induced 1 (RAI1). This condition affects at least 1 in 25,000 individuals worldwide. The major features of this condition include mild to moderate intellectual disability, delayed speech and language skills, distinctive facial features, sleep disturbances, dental and behavioral problems.This case report presents a 15 year old female patient with Smith-Magenis who was referred to Tufts University School of Dental Medicine(TUSDM) pediatric department for restoration of multiple carious lesions. The medical history reveals that patient had Ovarian cyst and ovary removed in 2020. The current medications are Zyrtec, Nasacort, Melatonin with no reported drug allergies. Clinical and radiographic examinations revealed brachyfacial face, concave profile and multiple carious teeth. This report will include clinical and radiographic findings associated with Smith-Magenis syndrome.

Introduction

- Smith-Magenis syndrome (SMS) is characterized by distinctive physical features (particularly coarse facial features that progress with age), developmental delay, cognitive impairment, behavioral abnormalities, self injurious behavior, sleep disturbance, and childhood-onset abdominal obesity [1]

PHYSICAL FEATURES
- Brachycephaly[1,3]
- Frontal bossing
- Hypertelorism
- Synophrys
- Upslanting palpebral fissures
- Midface hypoplasia[1,2,3]
- Broad square-shaped face with depressed nasal bridge
- Everted upper lip
- micrognathia in infancy
- relative prognathism with age

NEUROBEHAVORIAL FEATURES
- Infantile Hypotonia
- Sensory processing issues
- Developmental Delay
- Speech impairment
- Sleep Disturbance
- Inattention/ Hyperactivity
- Self Injurious behavior
- Impulsivity
- Attention seeking behavior
- Sensory processing issues
- Features of ASD
- REM sleep disturbance

Case Report

A 15 year old female patient with Smith-Magenis Syndrome was referred to TUSDM pediatric department for restoration of multiple carious lesions. The medical history reveals that patient had Ovarian cyst and ovary removed in 2020. The current medications are Zyrtec Nasacort, Melatonin with no reported drug allergies.
The patient has history of self injurious behavior.
The patient has sudden mood shifts and has explosive outburst and tantrums.
Extra oral Clinical examination revealed brachyfacial face, concave profile (Figure1), everted upper lip, depressed nasal bridge.(Figure 2)
Intra-Oral examination revealed that patient has Class I Molar relationship on Right (Figure4) and Left (Figure 5), lower midline is shifted 1mm to the right, open bite of 5mm, bilateral crossbite (Figure 3).
Radiographic assesment revealed dilaceration on tooth #20 and taurodontism on tooth #30.(Figure 8)

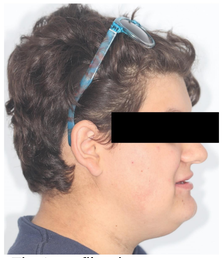

Fig.1 profile view. Concave profile, everted upper lip,

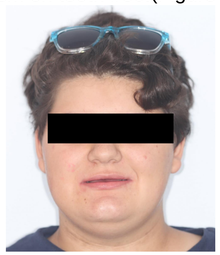

Fig.2 frontal view. Brachyfacial, depressed nasal bridge, hypertelorism

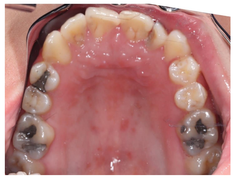

Fig.6 upper occlusal U shaped arch .

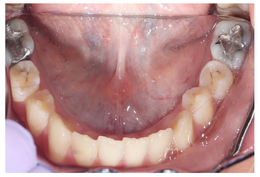

Fig.7 lower occlusal

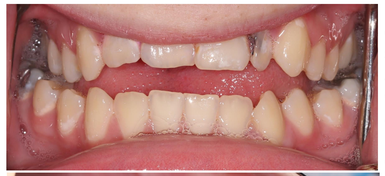

Fig.3 Intra oral frontal showing Anterior Open bite and posterior crossbite

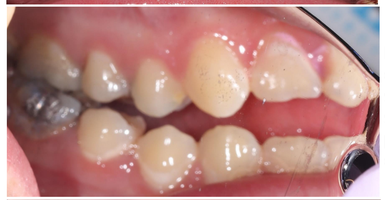

Fig.4 Intra oral right side showing posterior crossbite

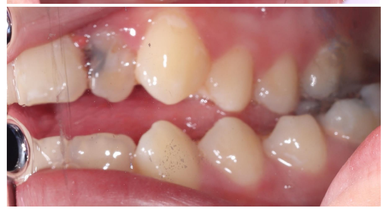

Fig.5 intra oral left side showing posterior crossbite

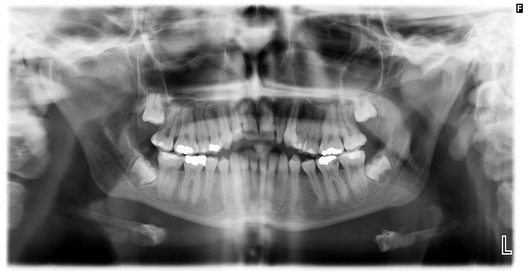

Fig.8 Panoramic Radiograph. Taurodontism #30, Dilaceration #20

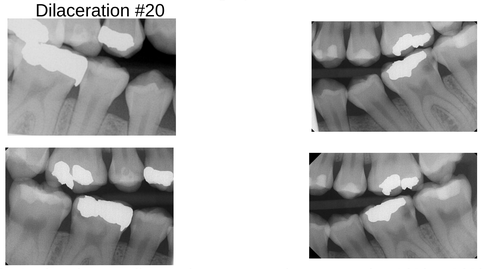

Fig.9 Bitewing Radiographs. Restoration on tooth #2(O), #3(O), #4(O), #5(DO), #11(O), #14(OL), #18(O), #19 (O) and #30 MOB)

Management

- Early morning appointments reccomended.
- Provided a stress free appointment.
- Short appointment as the patient is susceptible to mood swings.
- Restorations on tooth #2(O), #3(O), #4(O), #5(DO), #11(O), #14(OL), #18(O), #19 (O) and #30 (MOB) were completed using Nitrous Oxide Inhalation.
- Referred to OMFS Department for evaluation of 3rd molars
- Periodic Examination with prophylaxis and flouride varnish application every 3 months due to high caries risk.

Conclusion

To date, no specific therapy is available for Smith-Magenis syndrome; therefore, its clinical management consists of treating the medical issues presented by each affected individual. SMS patients have behavioral problems due to which their dental management may be challenging and pediatric dentists should be prepared to use different behavior guidance techniques.

References

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