DENTAL MANAGEMENT FOR A CHILD WITH SMITH-MAGENIS SYNDROME UNDER GENERAL ANESTHESIA: CASE REPORT

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Palavras-chave: Transtorno do Espectro Autista, Cárie Dentária, Genética, Síndrome de Smith-Magenis

RESUMO

Introdução: A síndrome de Smith-Magenis (SMS) é uma doença genética caracterizada por uma deficiência neuro-comportamental causada por mutações ou deleções no locus 17p11.2 compreendendo o gene 1 induzido por ácido retinóico (RAI1). O diagnóstico é feito por meio de análises clínicas em busca de características e para comprovar essa suspeita, é necessária a técnica denominada Hibridização In Situ por Fluorescência (FISH). **Objetivo**: O objetivo deste relato de caso é o primeiro a descrever o planejamento e execução do tratamento odontológico para uma paciente do sexo feminino de 5 anos de idade com SMS sob anestesia geral. **Relato do caso**: O paciente deu entrada no ambulatório da Universidade Federal Fluminense, com possível dor dentária, na anamnese observou-se a necessidade de tratamento invasivo em diversos elementos dentais e devido ao padrão de comportamento do paciente optou-se pelo tratamento sob anestesia geral. **Resultados**: Os procedimentos foram realizados (restaurações e extrações) no hospital na mesma etapa. O acompanhamento da criança após a intervenção foi a cada seis meses. **Conclusão**: A SMS é uma síndrome rara que requer amplo conhecimento do dentista e uma anamnese detalhada para a escolha da melhor opção para a solução do caso.

Keywords: Autism Spectrum Disorder. Dental Caries. Genetic. Smith-Magenis Syndrome.

ABSTRACT

Introduction: Smith-Magenis Syndrome (SMS) is a genetic disease characterized by a neuro-behavioral deficiency caused by mutations or deletions at the 17p11.2 locus comprising the retinoic acid-induced 1 (RAI1) gene. The diagnosis is made through clinical analysis looking for characteristics and to prove this suspicion, a technique called Fluorescence In Situ Hybridization (FISH) is required. **Objective**: The aim of this case report is to be the first to describe the planning and execution of dental treatment for a 5yearold female patient with SMS under general anesthesia. **Case report**: The patient was admitted to the clinic of the Universidade Federal Fluminense, with possible dental pain, in the anamnesis the need for invasive treatment was observed in many dental elements and due to the patient's behavioral pattern, treatment under general anesthesia was chosen. **Results**: Procedures were performed (restorations and extractions) in the hospital in the same step. The child follow-up after the intervention every six month. **Conclusion**: SMS is a rare syndrome that requires extensive knowledge of the dentist and a detailed anamnesis to choose the best option to solve the case.

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INTRODUCTION

The Smith-Magenis Syndrome (SMS) was first portrayed in the early 1980s.[1,2] It is considered a sporadic syndrome, showing a prevalence of approximately 1: 25,000 live births.[1,2] However, it is believed that this number is underestimated, many cases are not related due to lack of clinical knowledge for diagnosis.[3-7] The features of the syndrome begin to emerge from the 18th to the 36th month of life. Between the 2nd and 3rd year, behaviors typical of Autistic Spectrum Disorder (ASD) begin to emerge. Individuals with SMS usually have restricted interests, tendency to isolation and repetitive activities, characteristics that are similar to those of ASD. Although SMS is a differential diagnosis, the measures that are relevant for a child with ASD also benefit children with SMS.[5,8]

SMS is a neurological development disorder characterized by a well-defined pattern of abnormalities, including a distinct craniofacial dysmorphic model, abnormalities in the circadian rhythm of sleep-surveillance and cognitive impairment with behaviors and psychiatric disorders.[9] Craniofacial manifestations include brachycephaly; wide square face; synophrys; exaggerated eyelid lesions; low nasal bridge with a wide nasal base; everted upper lip and mandibular prognathism.[5] Some behavioral manifestations, such as self-injurious and angry outbursts, are common. In addition, most syndromes have mental retardation, ranging from mild to moderate.[9] Individuals with SMS demonstrate severe communication delays, which interfere with social interaction and learning.[10-13]

Craniofacial dysmorphisms can be observed, as well as an enlarged and prominent forehead, hypoplasia of the midline of the face, a prominent upper lip with the appearance of an arch and prognathism. In the oral cavity, some oral deficiencies are reported, such as lingual weakness, weak bilabial seal and abnormal palate. Although cleft lip and palate occur, they are less frequent in these individuals.[5,10]

Dental anomalies are usually found and can help in the diagnosis of SMS. Agenesis of one or more teeth, mainly of the lower second premolars, taurodontism and lacerations are the most frequently related. In intraoral examination, it is also possible to observe macroglossia and some oral habits.[10] Patients diagnosed with SMS with advancing age, commonly present tooth decay with the need for restoration procedures and gingivitis. This can be explained by the behavioral profile and the particularities of the SMS, which makes preventive care challenging.[10,14,15]

Depending on the physical, emotional and behavioral characteristics of the patients, outpatient dental care becomes unviable. In this situation, alternative methods are necessary, such as conscious sedation and general anesthesia, which is the option chosen in the present case.[16] This case report is the first to describe the planning and execution of dental treatment in a female patient which is a 5yearold child with SMS under general anesthesia.

CASE REPORT

Female patient, M.L.O.G., 5 years old, leukoderma, was taken to the patient with deficiency clinic of the Faculty of Dentistry of Universidade Federal Fluminense by the mother for the first dental treatment, with the main complaint that the daughter was possibly in pain and with large carious lesions, since she is not a verbal patient.

In the anamnesis, the mother reported that the daughter was diagnosed with Smith-Magenis Syndrome, Autism Spectrum Disorder and mental retardation. In Figure 1 the syndrome physical characteristics is observed frontal bossing, nasal and malar bone depression, and increased size of the bony chin. It was also reported that the patient is accompanied by a neurologist, psychologist, physiotherapist and speech therapist and regularly used medications such as Melatonin, Risperidone and Topiramate. During the interview, the mother commented that the daughter's diet included a bottle with sugary milk, twice a day. The mother also reported the difficulty of performing her daughter's oral hygiene.

At the clinical examination, it was observed that the child had a difficult behavior and was quite agitated for a thorough oral examination. However, it was possible to observe that the following dental elements had extensive caries lesions: 55, 54, 52, 51, 61, 62, 63, 64, 65, 75, 74, 84 and 85, and this can be observed in the panoramic radiography (Figure 2). For the panoramic radiography, the patient was medicated and cooperated. In view of the mother's complaint and the impossibility of treatment in the dental chair, the patient was scheduled to receive complete oral rehabilitation under general anesthesia. All preoperative exams were requested and thoroughly examined by the dental medical team, with special attention to the heart and renal disorders.

Extra and intra-oral hygiene was performed with 0.12% chlorhexidine and an pharyngeal pack was introduced. It was placed a lip retractor to better visualization. It was performed nasotracheal intubation and while the patient was sedated (Figure 3), dental prophylaxis was also performed with a low-speed handpiece and dental polishing brush. Subsequently, a restoration was carried out with composite resin A2 of the Z300 brand (A1 and B2 color, Resina Filtek Universal - 3M, São Paulo, Brazil) in the following elements: 55, 64, 65, 74, 84 and 85, with finishing and final polishing in the restorations. Due to severe caries and the

presence of an odontogenic abscess, seven teeth were extracted after administration of 3 tubes of 2% lidocaine with epinephrine 1: 100,000: 51, 52, 61, 62, 63, 54 and 75, with local suture Vicryl colorless (Ethicon, Johnson & Johnson, São Paulo, Brazil) thread (Figure 4). In the end, the pharyngeal pack was removed, the patient was extubated and sent to the room. The patient was discharged from the hospital on the same day. All guidance regarding the reduction of the cariogenic diet was given and oral hygiene instructions were given. The patient is followed up every six months. The patient has already attended two return visits. With integral restorations. Without presenting new caries lesions.



***Figure 1**: Facial and body (arms) characterization corresponding to SMS.*

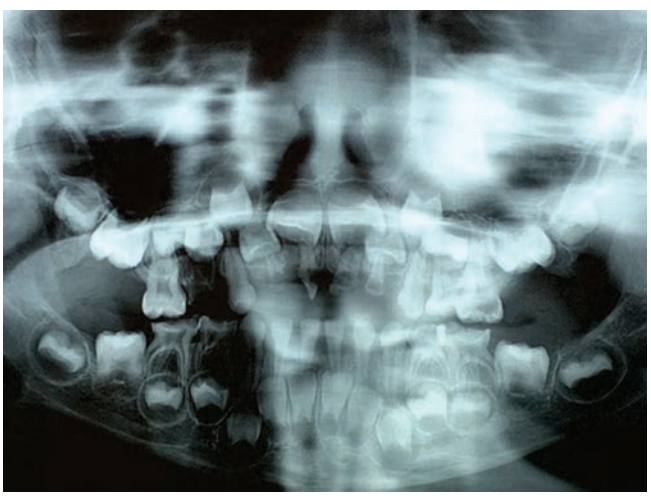


***Figure 2**: Dental anomalies observed in SMS. Taurodont pulps are present in upper and lower first permanent molars.*



***Figure 3**: Intubated patient. Intraoral preoperative photograph of the maxillary and mandibular arch showing tooth destruction, mainly upper lip.*

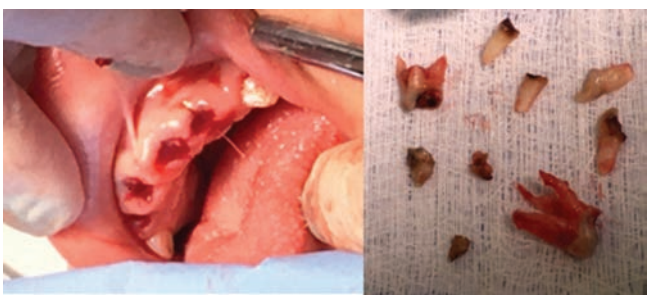


***Figure 4**: Intraoral postoperative photograph of the maxillary arch and teeth extracted. Observe the dental destruction of the elements.*

DISCUSSION

Smith-Magenis syndrome was first recorded by Smith et al. in 1982, and in 1986[1], it was further developed by Smith et al.[1] and Stratton et al.[2] Thus, it was characterized as a genetic disease that has the cognitive deficit caused by the deletion of the chromosomal region 17p11.2 or mutations of the RAI1 gene point.[1,2,7,17] However, there is little literature on the disease and reports of medical cases.[18] Reports of dental treatment and management of patients with SMS were not found in the literature.

This is the first work that reports the management and dental treatment of a child with SMS under general anesthesia. Report on the oral condition and its characteristics have little information and only one article was found.[10] The

consistent nding of anterior mandibular tooth position, frontal bossing, nasal and malar bone depression, and increased size of the bony chin was observed in this case (Figure 1 and 2), as well as Tomona and collaborators.[10] Another difficulty encountered was that the patient in this report was very young, with great difficulty in cooperating for outpatient care and in great need of dental treatment.

In the anamnesis, the mother reported that the patient was diagnosed as having the SMS and that she is monitored by a multidisciplinary team, using various medications. The diagnosis presented and the interaction with other professionals (doctors, nurses, speech therapists) brought an understanding of the clinical condition of this patient, making it possible to understand that due to the number of procedures to be performed, treatment in multiple sessions would not be indicated.[10,19]

Given the impossibility of effective behavior control within the dental clinic and the need for several applications and a large amount of dental procedures to be performed, with a lot of restorations and tooth extractions, the most accessible alternative was treatment using general anesthesia, thus performing all procedures in a safe and single session.

Dental treatment under general anesthesia should be done only when other methods of behavioral management are unsatisfactory, correctly adopting the previous measures and knowing the limitations of the technique.[20-22] Usually, it is indicated for patient with deficiency who have physical and mental restrictions, that is, unable to collaborate in cases of major interventions, making service in the office difficult.[23-26] For this, all necessary procedures were performed, such as: preoperative exams, surgical risk, scheduling of the surgical center at the hospital and request of an anesthesiologist for the application of general anesthesia.[22]

Currently known behavioral managements allow children and patients with deficiency to be facilitated, and a large portion of these patients can be seen on an outpatient basis using the different forms of non-pharmacological and pharmacological behavioral approach.[25-27] However, in extreme cases, the use of general anesthesia in the operating room becomes the most viable option for carrying out therapeutic strategies that favor the development of these individuals.[22] Education about oral hygiene should be intensive and should be given to the child and caregivers, as the child has involuntary movements and lacks the motor coordination necessary to brush properly independently. Special emphasis should be placed on the brushing technique; parental cooperation is extremely valuable in motivating and rewarding the child to brush properly. Constant and frequent compliments, when appropriate, is helpful. If parents are not motivated to become conscientious and compliant, and they do not maintain good home care, it is very difficult to keep the child free of caries, despite frequent dental consultations.

Future studies with groups of patients with SMS should be encouraged so that it is possible to know more deeply the oral characteristics and the management of these patients in view of the need for dental treatment. In addition, the multidisciplinary approach is important in order to allow patients to lead a better quality of life.

CONCLUSION

Smith-Magenis Syndrome stands out for being a rare syndrome and patients usually have difficult control during dental treatment. Thus, the need for a detailed treatment plan for choosing the most appropriate procedure is emphasized, aiming at a faster and more efficient intervention, preferably done in a single session.

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