Cognitive level, quality of life and oral health of a Prader-Willi Syndrome patient - case report with long-term follow-up

Nível cognitivo, qualidade de vida e saúde bucal de um paciente com síndrome de Prader-Willi - Relato de caso com acompanhamento de longo prazo

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ABSTRACT
Introduction: Prader-Willi syndrome (PWS) is a rare, neurobehavioral genetic disorder that alters the child's development. Concerning oral health, PWS patients present dental aspects that are not completely understood. Objectives: The aim of this case report was to describe long-term follow-up of a patient with PWS, regarding to cognitive level, overall self-perceived quality of life and oral health, sleep quality, anxiety trait, and oral condition. Methods: The information about his cognitive ability and socio-behavioral behavior was collected applying the questionnaires: Mini-Mental State Examination (MMSE), simplified World Health Organization Abbreviated Quality of Life survey (WHOQOL-bref), Sleep Assessment Questionnaire (SAQ), State Trait Anxiety Inventory (STAI-T) Oral Health Impact Profile (OHIP-14). The clinical examination was performed according to the WHO methods for oral health surveys. Index of Decayed, Missing or Filled teeth (DMFT) and other changes in mineralized tissues, such as dental fluorosis or erosion, incisor molar hypomineralization, enamel hypoplasia, bruxism, dental trauma and positioning of teeth in dental arches were also recorded. In the periodontal analysis, the visible plaque index, bleeding on probing and final diagnosis of gingivitis were performed. Data about dental occlusion and temporomandibular disorders were also noted. After data collection and clinical examination, a saliva sample was collected to assess salivary flow rate, pH and buffering capacity. Results: The analysis of the questionnaires revealed an individual satisfied with his quality of life, but very anxious. Clinical examination point to gingival inflammation located in the region of anterior teeth, maxillary and mandibular anterior tooth crowding and malocclusion. Salivary parameters were normal. Conclusion: This case report suggests that the PWS patient perceives that his quality of life is good despite his anxiety episodes. Regarding oral health, gingival inflammation and malocclusion are the main problems.

Keywords: Prader-Willi syndrome, Oral health, Saliva.

RESUMO
Introdução: A síndrome de Prader-Willi (PWS) é uma doença genética neurocomportamental rara que altera o desenvolvimento da criança. Relativamente à saúde oral, os doentes com PWS apresentam aspectos dentários que não são completamente compreendidos. Objectivos: O objectivo deste relatório de caso era descrever o seguimento a longo prazo de um paciente com PWS, no que respeita ao nível cognitivo, qualidade de vida e saúde oral em geral auto-percebida, qualidade do sono, traço de ansiedade, e condição oral. Métodos: A informação sobre a sua capacidade cognitiva e comportamento sociocomportamental foi recolhida aplicando os questionários: Mini-Mental State Examination (MMSE), inquérito simplificado sobre qualidade de vida abreviado da Organização Mundial de Saúde (WHOQOL-bref), Sleep Assessment Questionnaire (SAQ), State Trait Anxiety Inventory (STAI-T), Oral Health Impact Profile (OHIP-14). O exame clínico foi realizado de acordo com os métodos da OMS para os inquéritos de saúde oral. Foram também registados índices de dentes deteriorados, em falta ou cheios (DMFT) e outras alterações nos tecidos mineralizados, tais como fluorose dentária ou erosão,

hipomineralização dos molares incisivos, hipoplasia do esmalte, bruxismo, traumatismo dentário e posicionamento dos dentes nos arcos dentários. Na análise periodontal, foi realizado o índice de placa visível, sangramento na sondagem e diagnóstico final da gengivite. Foram também registados dados sobre oclusão dentária e distúrbios temporomandibulares. Após a recolha de dados e exame clínico, foi recolhida uma amostra de saliva para avaliar o fluxo salivar, pH e capacidade de tamponamento. Resultados: A análise dos questionários revelou um indivíduo satisfeito com a sua qualidade de vida, mas muito ansioso. O exame clínico aponta para inflamação gengival localizada na região dos dentes anteriores, apinhamento e má oclusão dos dentes anteriores maxilares e mandibulares. Os parâmetros salivares eram normais. Conclusão: Este relatório de caso sugere que o paciente PWS percebe que a sua qualidade de vida é boa, apesar dos seus episódios de ansiedade. Relativamente à saúde oral, a inflamação gengival e a má oclusão são os principais problemas.

Palavras-chave: Síndrome de Prader-Willi, Saúde oral, Saliva.

1 INTRODUCTION

Prader-Willi syndrome (PWS) is a rare, neurobehavioral genetic disorder that alters the child's development, affecting one child for every 10,000/30,000 births. It is a syndrome caused by the loss of activity of the paternal genes expressed in the proximal region of the long arm of chromosome 15, position q11-13 (Cassidy et al, 2012; Setti et al, 2012; Amaro et al, 2013), resulting in short stature, obesity, hypotonia, hyperphagia, sleep disorders, learning difficulties, psychiatric, endocrine disorders and cognitive impairment, which may increase the risk of developing oral disorders (Jin, 2011; Setti et al, 2012; Quaio et al, 2012, Deal et al, 2013).

Regarding systemic disorders, the syndrome is presumed to cause an important hypothalamic dysfunction, which decisively affects body homeostasis causing delayed in bone maturation, abnormal accumulation of fat (Miller et al, 2011; Amaro et al, 2013), cardiovascular and respiratory impairment, the latter being responsible for the high incidence of death among children and adults with PWS (Cassidy and Driscoll, 2009; Quaio et al, 2012; Amaro et al, 2013). Besides that, PWS individuals exhibit a specific characteristic behavioral that includes anxiety, cognitive rigidity, severe temper outbursts, obsessive-compulsive and self-injurious behaviors (Yang et al 2013; Rice et al, 2015).

Concerning to oral health, PWS individuals present dental aspects of interest that can be aggravated by their untimely behavior. Once long-term longitudinal description of oral PWS manifestations, as well as the aging process in PWS individuals are not completely understood, the aim of this case report was to describe a follow-up of nine years of a patient with PWS, regarding to cognitive level, self-perceived quality of life and of oral health, sleep quality, anxiety trait, and oral condition.

2 CASE REPORT

2.1. NON-CLINICAL DATA COLLECTION

The patient in this case report is the same previously described by Gadens et al (2014). The patient started his dental treatment at the PUCPR with 10 years old and in this study, we describe the oral condition after 10 years, that is, a 10-year long-term follow-up. At anamnesis and physical examination, the main marked physical alterations such as myopia, strabismus, short stature, maxillary lip, and labial commissures, facing down, small hands, and hypopigmentation of the hair, skin and eyes persist. Obesity seems to be the main problem, so much so that the patient underwent bariatric surgery.

To access information about their cognitive ability and their socio-behavioral behavior, the following diagnostic questionnaires were applied: a. Mini-Mental State Examination (MMSE): questionnaire to assess the individual's level of cognition and understanding of situations (Folstein et al. 1975); b. World Health Organization Abbreviated Quality of Life (WHOQOL-bref): used to assess the patient's overall quality of life (WHOQOL, 1998); c. Sleep Assessment Questionnaire (SAQ): self-assessment questionnaire for items with sensitivities and specificities for the six factors: insomnia, non-restorative sleep, disturbed sleep schedule, daytime sleepiness, sleep apnea and restlessness (Cesta et al., 1996); d. State Trait Anxiety Inventory (STAI-T): it is related to the person's personality and refers to differences in reaction to situations perceived as threatening with an increased state of anxiety (Spielberger et al, 1970) and e. Oral Health Impact Profile (OHIP-14): developed to assess the impact that oral health has on the quality of life of individuals, in a short-form, with 14 questions (Slade, 1997). The results of the questionnaires are collected and presented in Table 1.

Table 1. Main results of the applied questionnaires.

Questionnaire	Possible scores	Score achieved by the patient	Conclusion
MMSE	0-30	24	It allows to conclude that the patient's cognitive level is compatible with education equivalent to 4 years. It should be noted that in terms of attention and calculation, in which mathematical skills are required, the young man did not score, however in the other items, the score was extremely high.
WHOQOL-bref	0-5	4.5	Indicates that the patient perceives that his quality of life is good
SAQ	0-68	15	The score obtained indicates no sleep disturbance.
STAI-T	20-80	48	This finding means that the patient has moderate anxiety. It's a borderline value for severe anxiety
OHIP-14	0-56	3	The oral problems perceived by him did not impact the quality of life.

2.2 CLINICAL DATA COLLECTION

The clinical examination was performed by a researcher previously trained and was carried out according to the World Health Organization methods for oral health surveys (Petersen et al, 2013), and consisted of extra- and intra-oral examination where the face, teeth, and protective and support tissues were evaluated. Index of Decayed, Missing or Filled teeth (DMFT Index) and other changes in mineralized tissues, such as dental fluorosis or erosion, incisor molar hypomineralization, enamel hypoplasia, bruxism, dental trauma and positioning of teeth in dental arches were also noted. In the periodontal analysis, the plaque index, bleeding on probing and final diagnosis of gingivitis were performed according Caton et al (2018). Data about dental occlusion and temporomandibular disorders (TMD) were also recorded. Finally, it was noted the main dentistry necessity of the patient. The results of the clinical oral examination are shown in Table 2.

Table 2. Results of clinical parameters evaluated in the patient.

Clinical parameters			**Observations**
Mineralized dental tissues	Total teeth present	29	Radiographic examination revealed absence of teeth 28, 38 and 48. Two teeth were diagnosed with caries (26 and 27) and three teeth (16, 36 and 46) had restorations. Tooth 16 requires endodontic treatment. No other significant changes were observed in the mineralized structures of the teeth.
	Decay teeth	02	
	Missing teeth	00	
	Filled teeth	03	
	DMF-T index	05	
	Dental fluorosis	No	
	Dental erosion	No	
	Incisor molar hypomineralization	No	
	Enamel hypoplasia	Yes	
	Bruxism	No	
	Dental trauma	No	
Periodontal analysis	Visible plaque	Yes	Obtained data are indicative of gingival inflammation located in the region of anterior teeth, both in the maxilla and in the mandible.
	Bleeding on probing	Yes	
	Gengival inflammation	Yes	
	Periodontitis	No	
Dental oclusion	Anterior crossbite	No	The patient present significant alterations in the dental contacts due to inadequate dental position, especially in the anterior region, both mandible and the maxilla. Maybe the main therapeutic goal in this patient is restoring the static and functional occlusion through orthodontic treatment.
	Posterior crossbite	No	
	Maxillary anterior tooth crowding	Yes	
	Mandibular anterior tooth crowding	Yes	
	Angle classification	Class II, 1st division	
	Distal step	Yes, left and right	
	Overjet	8 mm	
	Overbite	5 mm	
	Canine guidance	Normal	
TMD	Myofascial pain	No	Despite the occlusal changes observed, the patient does not show signs or symptoms of TMD.
	Disk displacement	No	
	Arthralgia	No	

Note: The teeth were identified in this table according to the standards of the International Dental Federation.

Finally, after data collection and clinical examination, a saliva sample was collected to assess salivary flow rate, pH and buffering capacity. Regarding these parameters, both

salivary flow rate and pH showed values described in the literature for non-syndromic individuals (1.55 ml/min and pH = 7.9, respectively). The buffering capacity, that is, the ability of saliva to balance oral pH, was also considered normal.

3 DISCUSSION

In this case report it was described same aspects regarding quality of life, anxiety trait and oral condition of an individual with PWS. For that, validated questionnaires were applied to test cognitive level (MMSE), overall (WHOQOL-bref) and buccal (OHIP-14) quality of life, quality of sleep (SAQ) and traits of anxiety (STAI-T). Besides that, a nine years follow-up about buccal condition of this patient was performed.

It is not new that PWS is a genetic syndrome whose phenotype is characterized by neurobehavioral and cognitive disorders that can cause negative impacts on quality of life (Marsicano et al, 2011; Miller et al, 2011; Yang et al 2013; Rice et al, 2015), but the patient evaluated in this study revealed some interesting data that contradict this information. First, the applied cognitive test revealed a low score but the items that contributed to this low score were those that required mathematical skills. Only on these issues has performance been poor. Second, regarding quality of life, the questionnaires that evaluated this item indicate that the patient perceives that his quality of life is good, indicates that he does not have a sleep disorder and that oral problems do not impact his quality of life. Concerning to anxiety trait, the patient has a score that point to moderate anxiety, borderline to severe anxiety. During all dental appointments, these anxiety episodes were very rare and when they occurred, they were related to the dietary restriction to which the patient is submitted due to obesity. Most of the time, the patient was extremely friendly.

Maybe the main contribution of this work to the scientific literature is the long-term longitudinal description of oral manifestations in this patient since he has received dental care at the Clinic for Special Care Patients at the PUCPR for almost a decade. At that moment, the patient had complications caused by caries experience like missing teeth, residual roots, and caries in primary dentition. Now among the oral manifestations reported, dental caries and filled teeth were observed, but without missing teeth. A probable explanation for the high DMFT index is the hyperphagia presented by the individual. In fact, other studies also reported dental caries (Scardina et al, 2007; Roman-Torres et al, 2017) and, according to family members, it is usual for the patient to eat several times a day. No other significant changes, except for punctual enamel hypoplasia, were observed in the mineralized structures of the permanent teeth.

Concerning to periodontal condition, it is clear that there is a poor biofilm control. The visible plaque is present, and we detect bleeding on probing and gingival inflammation, however based in the diagnostic criteria, the patient does not have periodontal disease. Gingival inflammation is observed especially in the region of anterior teeth, both in the maxilla and in the mandible and we presume that is consequence of the tooth malposition and crowding plus poor biofilm control. Previous studies also reported high prevalence of periodontal pathology in an PWS individuals, and pointed out as primary etiology accumulation of biofilm, tooth malposition, and malocclusion (Yanagita et al, 2011; Hurren and Flack, 2016; Roman-Torres et al., 2017; Olczak-Kowalczyk et al., 2019). In fact, malocclusion seems to be a very prevalent condition in individuals with PWS. Bantim et al (2019) mentions references that cite malocclusion as a prevalent oral condition in these individuals. However, despite the occlusal changes observed, the patient does not show signs or symptoms of TMD.

Finally, saliva analysis was performed in order to test flow rate, pH and buffering capacity. Salivary flow rate, in contrast to what was observed nine years ago, was within the limits considered normal for individuals in this age group. This same patient in a previous case report expelled a very low volume of saliva characterizing hyposalivation. Buffering action and salivary pH were considered normal. Considering that, we presume that it is totally possible to maintain oral health in this individual but its necessary family involvement to instructed regarding to oral hygiene.

4 CONCLUSION

The data collected in this case report suggest that the PWS patient perceives that his quality of life is good despite his anxiety episodes. Regarding oral health, gingival inflammation and malocclusion are the main problems but, according to with patient's perception, did not impact his quality of life.

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CONFLICTS OF INTEREST

None.

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