*DEFB*1 c.-20G>A Variant in Mexican Patients with Early Childhood Caries

Flores Carrillo A[1], Ortiz Casillas MI[1], Mariscal Terrones J1 ,Chávez Maciel JM[1], Alvizo Rodríguez CR[2], Gutiérrez Angulo M[3], Beltrán Rivera M[4], Macías Montaño IA[4], Mariaud Schmidt RP[4]

1Posgrado de Odontopediatría, Departamento de Clínicas Odontológicas Integrales, Centro Universitario de Ciencias de la Salud, Universidad de Guadalajara. Guadalajara, Jal, México. 2 Facultad de Medicina, Benemérita Universidad Autónoma de Puebla. Puebla, México. 3 Departamento de Ciencias de la Salud. Centro Universitario de los Altos. Universidad de Guadalajara. Tepatitlán de Morelos, Jalisco. México.4Instituto de Investigación en Odontología, Departamento de Clínicas Odontológicas Integrales, Centro Universitario de Ciencias de la Salud, Universidad de Guadalajara. Guadalajara, Jal, México

Introduction

The American Academy of Pediatric Dentistry defines early childhood caries (ECC) as the presence of one or more surfaces with dental damage (with or without cavity lesion),missing (due to cavities) or filled surfaces on any deciduous tooth of a child between birth and 71 months of age (1).ICDAS II is a visual scoring system for caries detection, it describes six stages of caries severity, which varies from initial visible changes in the enamel to very noticeable cavitation in the dentin, allowing the recognition of the severity and incidence of cavities. (2).

Antimicrobial peptides (AMP's) are important contributors to maintaining the balance between health and disease and participate in processes such as modulation of the immune response, angiogenesis and cicatrization(3), these are classified into cathelicidins, histatins and defensins .Defensins are classified into α, β and θ subfamilies,(4) only α and β defensins are present in humans(5); β-defensin 1 is constitutively produced in parotid glands, buccal mucosa, tongue, gingival mucosa as well as in cells, ductal epithelial cells of minor salivary glands. As well a possess antibacterial activity against caries-related microorganisms and also play important roles in tooth pulp defense (6).Several author like Dale et al. (2006),(7) Ozturk et al. (2010)(8), Krasone et al. (2013) (9)Abbasoğlu et al (2014)(10),Yildiz et. al (2016)(11) and Ma. et al.(2023)(12) described the association between caries and c.-20G>A.

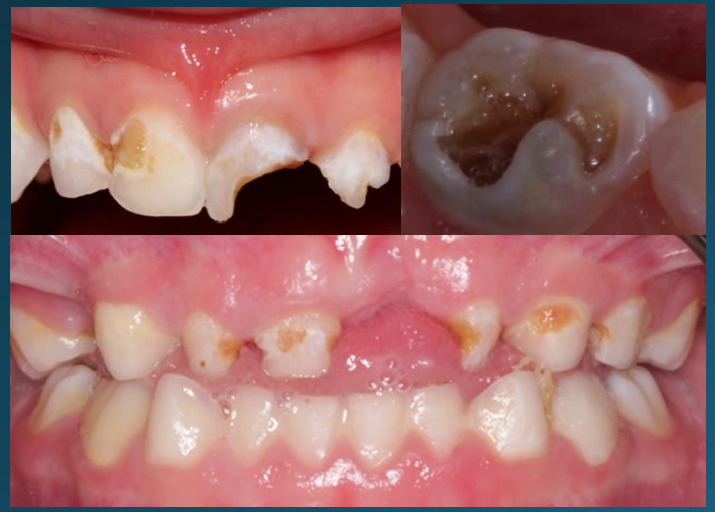

Figure 1. Clinical manifestations of early childhood caries: change in color to brown with loss of structure in the upper incisors and the lower molar.(own authorship).

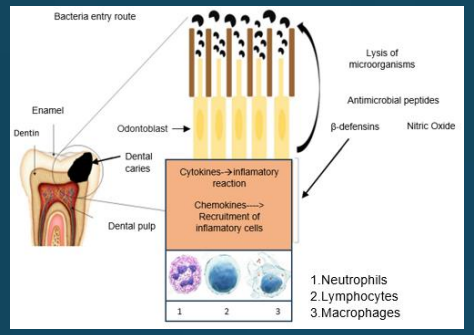

Figure 2. Role of human β-defensins in response to pathogens related to dental caries.(6) Modified by the author.

Aim.

Determine the association of the Defensin Beta 1 (DEFB1) c.-20G>A gene variant with ICDAS II in patients with early childhood caries.

Material and Methods

77 patients with a clinical diagnosis of early childhood caries and ages 6 to 71 months were recruited randomly, on their first appointment were asked with questions about the clinical history and assessment of the patients' general oral health status; All information regarding the development of the study was provided verbally to the father, mother or guardian of the potential participant, and a written survey was delivered to obtain personal data plus the consent letter under information.DNA was extracted from buccal mucosa cells. The c.-20G>A variant was identified by PCR-RFLP´s method. The association was estimated by odds ratio (OR) test and 95% of confidence Interval (CI).

Results

The age range of the patients was 6-71 months old. The patients in this study were separated in two groups according to ICDAS-II, the group I included 1-2 code (injury only in enamel) and was integrated by 13 ECC patients, while the group II included 3-6 code (injury found also in dentine) and were 64 patients. The DEFB1 AA genotype was more frequent in group II than I (39% and 15%, respectively). The comparison between both groups showed the AA genotype is associated with group II (ICDAS-II code 3-6) (OR=9.38, CI=1.14-74.84 and P=.02). Moreover, the DEFB1 A allele was also more frequent in group II than I (66% and 46% respectively) However, no significant differences were found for the A allele although the P value was in borderline (OR=2.31, CI=0 .99-5.42 and P=.05).

Genotypes *DEFB1* gene	Group I. ICDAS-II (Codes 1-2)	%	Group II. ICDAS-II (Codes 3-6)	%	P Value	OR (IC 95%)
GG	3	23	4	6		1 (reference)
GA	8	62	35	55	0.150	3.281 (IC=0.610-17.650)
AA	2	15	25	39	**0.018***	9.375 (IC=1.14--74.84)
Total	13	100	64	100		
Allele						
G	14	54	43	34		
A	12	46	85	66	**0.05****	2.31 (IC=.99-5.42)
Total	26	100	128	100		

Table 1. Genotypic and allelic distribution by study groups according to the ICDAS-II criteria.(13) Modified by the author.

Discussion.

The results obtained in the present study agree with those described by Yildiz, et al., 2015(11) in relation to the genotypic frequency, where the AA mutated genotype was 87.2% in the high-risk group and the A allele frequency was 68.9.% in the high-risk of caries in a Turkish adult population group. On the other hand, recently Ma. et al 2023 (12), in a study carried out on 1061 Chinese children, described that carriers of at least one mutated allele of the rs 11362 variant (CT or TT) who intake sugary foods more than once a week, had double risk of developing cavities. By the other hand, in 2018 Oliveira described that the mutated allele A confer double risk for the disease. This is the first study realized in the Mexican population, where the presence of early childhood caries was associated with the c.-20G>A variant of the DEFB1 gene (14); however, the data obtained in this research are relevant despite the sample size. Multiple authors have investigated the relationship of this variant with the caries and the results are controversial. This could be related with the origin population. Relating the ICDAS II index and the DEFB1 gene variant allowed us to broaden the panorama to provide an early diagnosis of susceptibility to caries and could be used as a tool for early detection of the disease.

Conclusion.

The AA genotype of DEFB1 c.-20G>A variant is a risk factor for early childhood caries development in Mexican patients.

References