

**Republic of Iraq
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College of Dentistry**



Assessment of CIITA rs4774 and rs6498122 polymorphisms in buccal swabs & blood samples of Iraqi oral lichen planus patients

A thesis

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Abstract

Background

Oral lichen planus is a common chronic inflammatory immune-mediated disease. The cause responsible for inducing oral lichen planus is still unidentified. Genetic cause has been proposed as etiologic factor. The class 2 transactivator gene has a great effect on autoimmune diseases. This gene is found on chromosome 16p13. In this chromosome there are many single nucleotide polymorphisms, two of them (rs4774 & rs6498122) have strong effect on autoimmune diseases. Studying the effect of these two single nucleotide polymorphisms has been hypothesized as predisposing factor in the pathogenesis of Oral Lichen Planus.

Aims of the study

This study aimed to:-

1-Study the effect of single nucleotide polymorphisms (rs4774, rs6498122) at class 2 transactivator gene by using buccal swabs and blood samples of oral lichen planus patients and compare their effect with healthy group.

2-Assess the validity of DNA extraction method from buccal swabs as convenient, painless and cost-effective alternative way for blood sampling for patients and researchers.

Subjects, Materials and Methods

This study was performed on 30 buccal swab and 30 blood samples from patients with oral lichen planus, and 30 buccal swab and 30 blood samples from healthy people as control. DNA was extracted from these samples, primer and probe were designed especially for (rs4774 & rs6498122) and master mix in quantitative Real time Polymerase Chain Reaction machine are used.

Results

It has been found that oral lichen planus cases were among females which representing 73.3% of the total number, while among males were 26.6% . The most frequent clinical form of oral lichen planus was the reticular type 86.6 % ,while each of erosive type and reticular type with melanin pigmentation represented 6.66% of the total cases.

Similar result found for both buccal swabs and blood samples regarding single nucleotide polymorphism for both rs4774 & rs6498122.

The single nucleotide polymorphism rs4774 had a significant difference on Hetero genotype, $P = 0.01^*$, and non-significant difference of Mutant genotype , $P = 0.11$, and the mutant allele C had a very high significant difference on this single nucleotide polymorphism , $P = 0.0001^{***}$.

The single nucleotide polymorphism rs6498122 had a non-significant difference on Hetero genotype, $P = 0.514$, and very high significant difference of Mutant genotype , $P = 0.0009^{***}$, and the mutant allele G had a very high significant difference on this single nucleotide polymorphism , P value $= 0.0001^{***}$.

When the two single nucleotide polymorphisms were found in the same patient (combination method) , significant difference has been found in patients with Wild/Mutant genotype for SNPs rs4774/rs6498122 ($P = 0.002^{**}$ with OR=zero) , as well as in patients with Hetero/Hetero genotype ($P = 0.015^{**}$ Odd Ratio=3.5 CI% =0.65-18.98).

Conclusions

Data showed that the two single nucleotide polymorphisms rs4774 & rs6498122 at class 2 transactivator gene are associated with oral lichen planus and could also indicate the autoimmune characteristics of oral lichen planus . Buccal swab can be used instead of blood sample in single nucleotide polymorphisms genetic research .