



## Mutation Heteroplasmy of One Nucleotide Position in Iraq population

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### Summary

Detectable heteroplasmy can be observed as point heteroplasmy where two DNA bases are observed at the same nucleotide position. The aim of this study is to search on the mtDNA heteroplasmy which may be useful in forensic genetics applications and determining the Iraqi population history. Mitochondrial DNA hypervariable regions I and II of control region were sequenced from 100 random healthy unrelated individuals of three sequential generations belong to the Arab ethnic of Iraqi population. The results showed that point heteroplasmy at position 16233 record haplotype 16233Y 16290T 16319A 16362C 73G 235G 309.1C which classified to haplogroup A4.

**Key words:** mitochondrial DNA, heteroplasmy, DNA Sequencing, Iraqi population.

### Introduction

Heteroplasmy is defined as the presence of more than one type of mtDNA sequence in an individual; that heteroplasmy can be expected to play a role in forensic interpretation on a regular basis, and that knowledge of its biological foundations contribute to conservative, and scientifically appropriate interpretational guidelines (1). A phenomenon which is referred to as heteroplasmy exists in mitochondrial DNA where a single individual carries two different populations of mitochondrial DNA either in a tissue, cell, or in the mitochondrion. Heteroplasmy can improve the probability of a match. The first sequence heteroplasmy accounted in the forensic identification

involved the identification of the Russian Tsar Nicholas II (2, 3). The mtDNA sequence of the Tsar Nicholas II putative remains was found to match the sequence of his living maternal descent except at one particular sequence. Tsar Nicholas II was found to have both T and C nucleotides at position 16, 169, that confirm the remains were belonged to Tsar Nicholas II, his late brother Grand Duke of Russia Georgij Romanov which was exhumed and tested with the same procedure. His mtDNA sequence was found to be identical with Tsar Nicholas II, including bearing the same heteroplasmy at position 16, 169. Since the extreme rarity of this heteroplasmy to happen by chance between unrelated

individuals, the remains was declared as Tsar Nicholas II and put to rest with

### Materials and Methods

The present study was randomly selected hundred blood samples of unrelated Iraqi volunteers of three sequential generations. Mitochondrial DNA was isolated by using mtDNA extraction Kit (Biovision, USA). PCR amplification of the mtDNA D-loop region generates four partially overlapping PCR amplicons (~350 bp for each one) spanning the four hypervariable fragments ranging from 15997-

respect (3).

16236, 16159-16527, 29-285 and 172- 419 nucleotide position in the mtDNA D-loop; the four amplified products were subjected to cycle sequencing by using ABI 3730 xL DNA analyzer (Applied Biosystem, USA). The DNA Sequence Data were analyzed by using BioEdit Software and then aligned with the revised Cambridge Reference Sequence (GenBank sequence NC\_012920) by added the CLUSTAL W to the same software.

### Results and discussion

One point mutation heteroplasmy was detected in Arab Iraqi sample by direct sequencing of the control region fragments. It was located at position 16233 that showed both nucleotide Y= C/T at this position as shown in figure (1). Heteroplastic T and C at this specific position is not able to be detected by direct sequencing, in spite of the quality score of this position which approved confirmatory results of this heteroplasmic point mutation the light and heavy strand examination was carried out.

In general, the heteroplasmy is considered a disadvantage in forensic casework, it can complicate and invalidate data interpretation, but in this study, that sample at position 16233 record haplotype 16233Y 16290T 16319A 16362C 73G 235G 309.1C which classified to haplogroup A4, whereas in the study of Al-Zahery, the results showed position 16233C in Arab Iraqi case, which recorded mtDNA haplotype T2e. This special feature help improved data interpretation and probability of match (4).

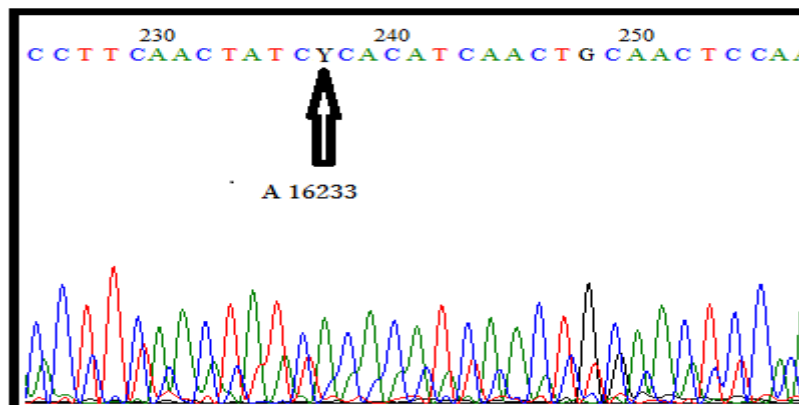


Figure (1): Electropherogram DNA sequencing of sample (A04) showing Heteroplasmy nucleotide position 16233 by Bioedit 7.2.5 software.

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## طفرة هتيروبلازمي عن موقع نيوكليوتيدة واحدة في المجتمع العراقي

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## الخلاصة:

يمكن تحديد الهتروبلازمي من ملاحظة الهتروبلازمي النقطية في قاعدتين الدنا في موقع النيوكليوتيدات نفسه. الهدف من هذه الدراسة هو البحث على الهتروبلازمي في الدنا الماييتوكونديري والتي قد تكون مفيدة في تطبيقات الوراثة الجنائية وتحديد تاريخ المجتمع العراقي. المناطق فائقة التغير للدنا الماييتوكونديري الأولى والثانية من منطقة التحكم ، اجري تتابع الدنا من 100 شخص ليسوا لهم صلة قرابة لثلاثة أجيال متتابعة وبشكل عشوائي وتنتمي إلى العرق العربي العراقي. أظهرت النتائج أن الهتروبلازمي النقطية في الموقع 16233 سجل هابلوتايب 16233Y 16290T 16319A 16362C 73G 235G 309.1C التي تصنف إلى هابلوغروب A4.