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The genetic male legacy from El Salvador

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Abstract

Allele frequencies and haplotype and haplogroup analysis have been performed for 16 Y-chromosome binary markers and 8 Y-chromosome STRs (DYS19, DYS385I and II, DYS389I and II, DYS390, DYS391, DYS392, DYS393). Data was obtained from a general sample of 93 unrelated individuals living in metropolitan areas from El Salvador, and 67 individuals from different historical ethnic groups, Conchagua, San Alejo, Panchimalco, Izalco and finally Nueva Concepción with white people. Levels of admixture among metropolitan and rural areas were evaluated and population substructure measured.

A total of 13 haplogroups and 136 different haplotypes were found. The most frequent haplogroup in the general metropolitan population was the European R1b, while in the Indigenous samples considered as a whole the most frequent was the Amerindian haplogroup Q3.

Keywords: Y-chromosome; STRs; SNPs; El Salvador; Metropolitan populations; Indigenous groups

1. Introduction

Haplotypes based on genetic markers of the non-recombining part of the Y-chromosome are becoming an important tool for the forensic investigation [1,2].

Because of the extremely high diversity of Y-chromosome haplotypes in most of the populations and because the product role cannot be used to estimate Y-STR haplotype frequencies, extensive databases are needed and therefore, they are been performed. The first free online database created was the Y-haplotype reference database (YHRD, http://www.yhrd.org), including the established standards of minimal and extended haplotypes.

In the case of Y-chromosome binary polymorphisms, based on studies of global populations there is extensive knowledge of its high geographic specificity. SNP haplogroups can be used to estimate admixture levels among populations and to evaluate population stratifications [2].

El Salvador is the smallest country of Centro America (21,000 km^2), but it is the most densely populated of Latin America. It has approximately 6.2 millions inhabitants. Near 90% of the population is mixed, between Europeans and Indigenous, and only 1% are Indigenous people. Metropolitan area from San Salvador represents 1.7 million people, and approximately 42% of the population lives in rural areas [3].

In this study general population from metropolitan areas was compared with Indigenous samples scattered in four different rural areas, Conchagua, San Alejo, Izalco and Panchimalco. In addition some white individuals from the rural area of Nueva Conception were also included.

Population from Conchagua lived in the islands of Fonseca's Gulf, which is shared nowadays by El Salvador, Honduras and Nicaragua. In 1522 it was discovered by a Spanish expedition. The Spaniards and the English pirate invasions caused the ''Lencas'' emigration towards firm land, the place in which, now, the natives are settled down in the eastern area of El Salvador.

San Alejo is a very successful land (in its best moment) placed in the eastern area of El Salvador, which was inhabited by ''Lencas''. It was colonized by Spaniards in the XVI century.

The Izalco population originating from Yaqui or ''Pipil'' is placed in the western area of El Salvador. It was one of the most important native settlements of the pre-Columbian period whose beginnings date back to the second half of XI century after Christ. It was conquered by Spaniards in the first half of the XVI century.

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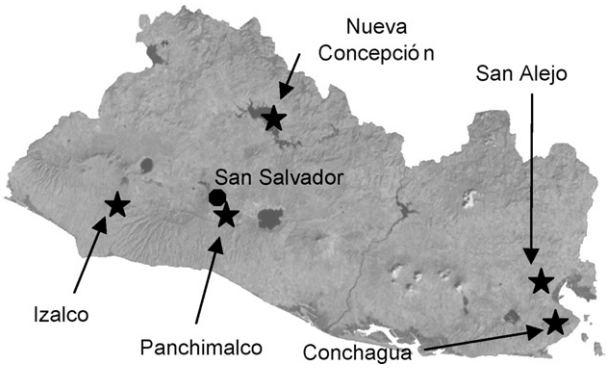

Fig. 1. Map of El Salvador. Stars represent the location of the populations analysed.

Panchimalco is also a population originated from pre-Columbian period. It is placed in the Central area of El Salvador, established by the ''Pipil'' emigrants as a result of the dispersion of the different tribes ''Nahoas'' during the XI and XII centuries after Christ. In the first decades of the XVI century it was also colonized by Spaniards.

In this study, we examine the male lineage structure of the population from El Salvador, by means of analysing the highly variable Y-STR haplotypes and the geographically structured Y-chromosome haplogroups. Levels of admixture among metropolitan and rural areas were evaluated and population substructure measured.

2. Materials and methods

2.1. DNA samples

A total of 187 males with different geographic origin from El Salvador have been analysed (Fig. 1). The selected samples include 93 individuals from a general population from El Salvador and 67 individuals from four historical ethnic groups (Fig. 1): Conchagua ($n = 23$), San Alejo ($n = 12$), Panchimalco ($n = 11$), Izalco ($n = 12$) and finally Nueva Conception ($n = 9$) with non-Indigenous people living in rural areas. Although the number of samples for some of the population groups is quite reduced, we decided to send this data to publication anyway, because of the high difficulty in getting additional samples.

DNA was extracted from blood stains using a standard phenol–chloroform method. DNA quantification was performed using fluorescence detection with a DyNAQuant 200 (Amersham Biosciences, Uppsala, Sweden).

2.2. Y-chromosome SNP analysis

We typed 16 Y-chromosome binary markers (Fig. 2): SRY-1532, M213, M201, M170, M26, 12f2,1, M62, M172, M9, M70, M22, Tat (or M46), 92R7, M3, M173 and P25. Conditions described by Brión et al. [4] were followed to analyse nine of the SNPs in a multiplex 1, and six markers in a multiplex 2. The remaining polymorphism, M3, was analysed in a single PCR reaction using 200 μM of each dNTP, 1.5 mM of $MgCl_2$, 0.5 U of taq DNA polymerase, and 0.25 μM of each of the following primers F: CTGCCAGGGCTTTCAAATAG and R: AAGGGCATCTTTCATTTTAGGT. Cycling conditions were 30 cycles of 94°30″, 57°1′, 72°1′. Detection was performed by single base extension, with the SNaPshot multiplex kit (Applied Biosystems). The probe used for the detection reaction was GGGTCACCTCTGGGACTGA.

Haplogroups were named according to the proposals of the Y-Chromosome Consortium [5,2].

2.3. Y-chromosome STR analysis

The eight Y-STRs included in the European minimal haplotype described in the Y-STR haplotype reference database (http://www.ystr.org) were analysed in the whole sample. In addition the samples belonging to the Indigenous ethnic groups were also analysed for the DYS437, DYS438 and DYS439.

In the case of the general population from El Salvador PCR reactions and detection were performed as previously described in Lovo et al. [6]. STR analysis in the Indigenous samples was performed using the commercial kit Powerplex® Y System (Promega Corporation), following the recommendations of the manufacturer. Products were run on an ABI 3100 Genetic Analyser (Applied Biosystems) and analysed using the GeneScan 3.7 or the Genotyper 3.7 software (Applied Biosystems).

Allele designation for the STRs was performed according to Gill et al. [7] using allelic ladders available with the kit.

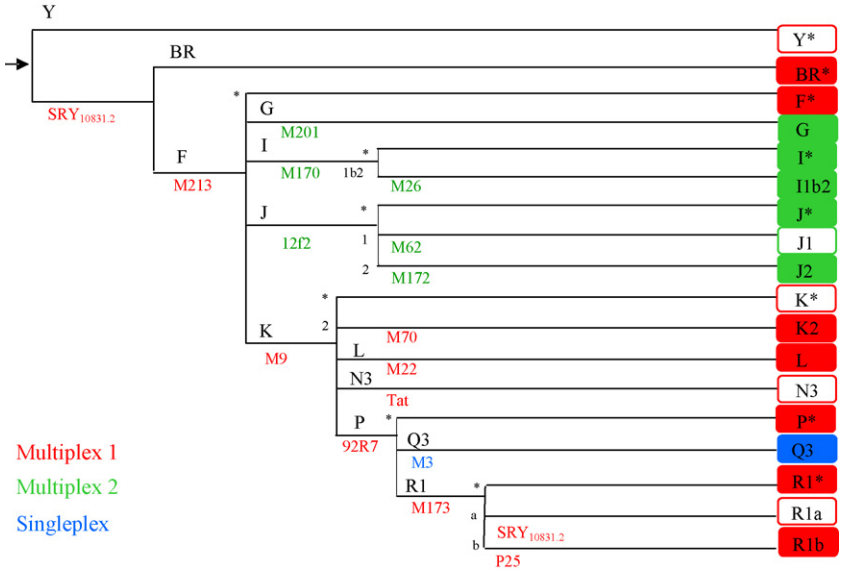

Fig. 2. Maximum-parsimony tree of 18 Y-chromosome haplogroups. Major clades are labelled with large capital letters above each branch. Mutations names are given below the branches. The length of each branch is not proportional to the age of the mutation. Different colours represent different amplification and detection reaction. Haplogroups found in our study appear with coloured squares. (For interpretation of the references to colour in this figure legend, the reader is referred to the web version of the article.)