Deletion mapping and paternal origin of a Mexican AMELY negative male

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ABSTRACT

AMELY deletion mapping represents the gender marker most widely used for human identification and biomedical purposes. However, some failures in sex-typing have been observed globally. In this study, we could approximate the population frequency of AMELY negative males in 1230 individuals from five states of Mexico (0.0812). For the sole AMELY negative male detected, we constructed a deletion map by means of 10 markers (2 STRs and 1 Y-STR). This allowed classifying the case into the most common category (Class I deletion), according to the nomenclature proposed by Jobling et al. (2007). Interestingly, the Mexican sample was R1a1, a Y-chromosome haplogroup non-previously reported for AMELY negative cases. The geographic distribution of R1a1, and the Y-STR haplotype similarity with a reported case from Slovenia, suggests an Eastern-European paternal origin for this case from Mexico. To our knowledge, this is the first report in Latin America that implies a low population frequency and European paternal origin of AMELY negative cases.

1. Introduction

The AMELY gene probably represents the marker most widely employed for gender determination, human identification and biomedical purposes. Several commercial multiplex PCR kits used in forensic genetics have incorporated this locus to establish the gender of biological samples, which has primary importance during the investigation process. Nevertheless, some failures in gender typing have been observed globally [1–3]. Some authors have argued that this "sex-typing error" is due to a rare mutation in the AMELY gene [2], whose frequency is higher in Indian groups than in other populations [4]. The incorrect sex designation of a male as "female" has serious consequences in prenatal diagnosis of X-linked diseases, and this represents confusing evidence in forensic cases. Because of the lack of clinical manifestations, we can predict the frequency AMELY negative males is underestimated, and the real impact for human identification purposes is unknown in the majority of populations. Although ideally the information of AMELY negative individuals could be improved including data from paternity and forensic cases without additional cost, probably they are not reported because they do not constitute a relevant fact, either for the study's conclusion or for the laboratory's interests. This fact justifies carrying out straightforward analysis concerning the structural variation and population distribution of these cases. Presently, the largest study included 45 AMELY deletion males from 12 Asian and European populations, predominating the Y-chromosome haplogroups J2e1 and R1b1b, respectively [5]. Although this effort allowed proposing a nomenclature of five deletion classes (I, II, III, and IV), eventually one AMELY deletion pattern that does not correspond with the cited nomenclature was reported [6].

Respect to AMELY negative males in Latin America, we did not find previous reports concerning the population frequency and structural characterization. This is interesting because admixed individuals, also described as mestizos, have peculiar genetic admixture patterns including different proportions of European, Amerindian, and African components. Particularly in Mexico, significant differences of these admixture proportions have been inferred by means of autosomal short tandem repeats (STRs) and Y-linked STRs (Y-STRs) commonly employed for human identification purposes [7,8]. In this study we approximate the population frequency of AMELY negative males in a Mexican population sample including five states. Additionally, we described the deletion map and ancestral origin of the unique case detected by means of autosomal STRs, Y-STRs, and Y-linked single nucleotide polymorphisms (Y-SNPs).

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