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Research paper

Extensive geographical and social structure in the paternal lineages of Saudi Arabia revealed by analysis of 27 Y-STRs

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ABSTRACT

Saudi Arabia's indigenous population is organized into patrilineal descent groups, but to date, little has been done to characterize its population structure, in particular with respect to the male-specific region of the Y chromosome. We have used the 27-STR Yfiler® Plus kit to generate haplotypes in 597 unrelated Saudi males, classified into five geographical regions (North, South, Central, East and West). Overall, Yfiler® Plus provides a good discrimination capacity of 95.3%, but this is greatly reduced (74.7%) when considering the reduced Yfiler® set of 17 Y-STRs, justifying the use of the expanded set of markers in this population. Comparison of the five geographical divisions reveals striking differences, with low diversity and similar haplotype spectra in the Central and Northern regions, and high diversity and similar haplotype spectra in the East and West. These patterns likely reflect the geographical isolation of the desert heartland of the peninsula, and the proximity to the sea of the Eastern and Western areas, and consequent historical immigration. We predicted haplogroups from Y-STR haplotypes, testing the performance of prediction by using a large independent set of Saudi Arabian Y-STR + Y-SNP data. Prediction indicated predominance (71%) of haplogroup J1, which was significantly more common in Central, Northern and Southern groups than in East and West, and formed a star-like expansion cluster in a median-joining network with an estimated age of ~2800 years. Most of our 597 participants were sampled within Saudi Arabia itself, but ~16% were sampled in the UK. Despite matching these two groups by home sub-region, we observed significant differences in haplotype and predicted haplogroup constitutions overall, and for most sub-regions individually. This suggests social structure influencing the probability of leaving Saudi Arabia, correlated with different Y-chromosome compositions. The UK-recruited sample is an inappropriate proxy for Saudi Arabia generally, and caution is needed when considering expatriate groups as representative of country of origin. Our study shows the importance of geographical and social structuring that may affect the utility of forensic databases and the interpretation of Y-STR profiles.

1. Introduction

Saudi Arabia is the largest country in the Arabian Peninsula. Its population of ~32 million people is distributed highly non-uniformly (Fig. 1), with very low densities in its large desert areas, but high densities concentrated around a small number of cities. Its indigenous Arab people (~63% of the population; www.stats.gov.sa, accessed 12/07/17) are historically organized into geographically-differentiated patrilineal descent groups, or tribes [1], with a tradition of consanguinity [2]. This geographical and social organization might be expected to have an effect on patterns of genetic diversity, particularly regarding the male-specific region of the Y chromosome (MSY), which in turn could have implications in interpretation of DNA profiles.

Genetic studies on Saudi Arabia to date are limited. Exome sequencing of a set of samples from the Arabian Peninsula including Saudi individuals demonstrated relatively high inbreeding coefficients [3], consistent with a history of consanguineous marriage. A general analysis of Saudi Arabian mitochondrial DNA (mtDNA) diversity [4] showed a pattern of haplogroups similar to that of other Arabian Peninsula samples. In another mtDNA-based study [5] – the only example to divide Saudi Arabia sub-regionally – central, northern, western and southeastern sub-groups formed a single cluster in a multi-dimensional scaling (MDS) analysis when compared to other Arabian Peninsula samples, but also presented significant inter-group differences. Y-chromosome studies have analysed the seven Y-STRs defining the minimal haplotype [6], or haplogroup-defining SNPs together with 17 Y-STRs (Yfiler®) for one specific haplogroup [7]. The first of these [6] revealed lower diversity in Saudi Arabia than in populations from

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Fig. 1. Map of Saudi Arabia, showing population density and sub-regional divisions used in this study. Population density is indicated by shading as shown in the key, top right, and locations of some major cities are shown. Adapted from Global Rural-Urban Mapping Project (sedac.ciesin.columbia.edu/gpw/), under a Creative Commons 3.0 Attribution License. Administratively, Saudi Arabia is divided into 13 regions which we here consider as five larger geographical areas, namely: Central (Riyadh, Al-Qassim), Northern (Northern borders region, Tabuk, Al-Jawf and Hail), Southern (Asir, Jazan, Bahah and Najran), Eastern (Eastern province) and Western (Mecca and Medina).

outside the Arabian Peninsula, and affinity between Saudi Arabia and Yemen, which together were strongly differentiated from Oman and Dubai. It was speculated that this might be due to the influence of patrilineal descent and polygyny. The second study [7] showed that haplogroup J1 was the most prominent lineage (42%) in the Saudi Arabian sample studied, and that genetic distances based on haplogroup frequencies were relatively small among Arabian Peninsula samples. The focus of Y-STR typing on one lineage precludes any population-based conclusions on haplotype diversity from this study.

To date, therefore, while some general studies have been carried out, little has been done to characterize population structure within Saudi Arabia. Knowledge of any such structure is important in the interpretation of the significance of DNA-based forensic evidence, and in the construction of appropriate databases. Here, we use the 27 Y-chromosomal short-tandem repeats (Y-STRs) in the Yfiler® Plus kit to characterize haplotypes in 597 Saudi males sub-divided by geographical region. We consider the relationships of Y-chromosome diversity between regions within the country and also between Saudi Arabia and other surrounding populations. Finally, we compare the spectrum of Y-chromosome types in males recruited within Saudi Arabia with that of regionally-matched males recruited in the United Kingdom, to ask if social structuring also influences patterns of Y-haplotype diversity.

2. Materials and methods

2.1. DNA sampling

Five hundred and ninety-seven DNA samples were collected from indigenous Saudi Arabian males who were ethnically and linguistically Arabic. Of these, 503 were extracted from blood spots on FTA cards (Whatman, UK), sampled from individuals recruited within Saudi Arabia itself. The remaining 94 were extracted from buccal swabs [8], or from saliva samples via the Oragene kit (DNA Genotek), from Saudi males resident within the UK. In each case, males with ancestry (to the level of paternal great-grandfather) from five geographical subdivisions of the country shown in Fig. 1 (Central, Northern, Southern, Eastern, and Western) were sampled, and consideration of relatedness ensured that all sampled males were separated by at least three generations. Ethical review for recruitment and analysis was provided by the Saudi General Administration for Forensic Evidence and the University of Leicester Research Ethics Committee. Informed consent was provided by all participants.

2.2. DNA extraction and quantification

DNAs were extracted and purified from FTA blood-spot samples using a fully automated STARlet workstation (Hamilton) and the PrepFiler® Forensic DNA Extraction Kit (Thermo Fisher Scientific), starting from 1.2-mm diameter punches produced using the BSD100 Punching System (Microelectronic Systems). Buccal samples were extracted via QIAamp DNA Mini Kits on a QIAcube robotic workstation (Qiagen). All DNA samples were quantified using the Quantifiler® Human DNA Quantification Kit (Thermo Fisher Scientific) on an Applied Biosystems® 7500 Real-Time PCR System.

2.3. DNA amplification and fragment detection

The Yfiler® Plus PCR Amplification Kit was used to generate Y-chromosome haplotypes for the 27 STRs DYS19, DYS385a, DYS385b, DYF387S1a, DYF387S1b, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS449, DYS456, DYS458, DYS460, DYS481, DYS518, DYS533, DYS570, DYS576, DYS627, DYS635, and Y-GATA H4. PCRs were conducted as recommended by the manufacturer on a Veriti (Thermo Fisher Scientific). Fragments were detected using an ABI3500 or ABI3130xl

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