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Maternal and paternal contributions in Jewish populations

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Abstract. A total of 107 Jewish individuals (Ashkenazi, Sephardic, North African and Oriental) have been analyzed for the Y-chromosome minimal STR-haplotype and for the HVRI mtDNA. The results have been compared with autosomal microsatellite data obtained previously from the same population samples. Significative sex-specific differences could be observed in these Jewish populations.

Keywords: MtDNA (HVRI); Y-chromosome STRs; Jewish populations

1. Introduction

Modern Jews constitute one ethnic group split into several groups, the most numerous of which are the Ashkenazim, who have resided in north-eastern Europe for centuries; the Sephardim who, after their expulsion from Spain in 1492, lived in other Mediterranean countries, especially Turkey; the North African, where evidence exists of Jewish communities as early as the first centuries AD that were augmented as a consequence of the Spanish expulsion; and the Oriental Jews who have lived in Middle East countries throughout their history [1]. Geneticists have studied Jewish populations since the turn of the 20th century in an attempt to unravel what must be a complex system of interrelationships among Jewish communities and their non-Jewish neighbours. Several studies of ''classical'' and DNA markers have provided evidence both for the common genetic origin of Jewish communities and for admixture between Jewish communities and their host peoples. Studies based on low-resolution mtDNA RFLP or HVRI sequences have been published [2,3] but no mtDNA databases have been reported to date for Jewish populations, which are essential in order to estimate random match probabilities in forensic casework. Respect to Y-chromosome, there is also a lack of database containing the minimal Y-chromosome haplotype.

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The purpose of the present study were: (i) to develop a HVRI sequence mtDNA database from Jewish individuals, (ii) to study the Y-chromosome "minimal haplotype" in these populations and, (iii) to compare the differences in mtDNA found in Jews and non-Jews with those found using Y-chromosome and autosomal microsatellite data obtained from the same population samples.

2. Material and methods

We analyzed 107 Jewish individuals. Following the classical criteria, they were categorized in four groups: Ashkenazi, Sephardic, North African and Oriental. All these samples belong to the collection of The National Laboratory for the Genetics of Israeli Populations at Tel-Aviv University.

A 406-nt sequence in region I of the mtDNA D-loop (sites 15994–16400) was analysed. Amplification was performed on a 2400 GeneAmp Thermal Cycler (Perkin Elmer). A QIAquick PCR purification kit (Quiagen, Germany) and the Big Dye Terminator Ready Reaction kit (Perkin Elmer) was used. Sequencing reaction products were analysed using an ABI PRISM 310 (PE/ABD) automated DNA sequencer. Each template was sequenced in both directions.

The five Y-chromosome STRs DYS19, DYS389I and II, DYS390 and DYS391 were co-amplificated. PCR amplification of DYS393 was achieved in singleplex and coamplification of the loci DYS385 and DYS392 was performed in a duplex reaction. A GeneAmp PCR System 2400 and an ABI 310 automatic sequencer (PE Applied Biosystems) was used.

3. Results and discussion

In mtDNA a total of 74 different haplotypes in 107 individuals were observed, with a lower incidence of unique haplotypes (54.2%) than in other populations (Table 1). When compared to the CRS sequence, 75 transitions, seven transversions and one insertion were found. Three individuals with one position of sequence heteroplasmy at nucleotides 16093, 16134 and 16169, respectively, were detected. The mean pairwise differences in the Jewish populations were in the range of 5.5–6.2, with the exception of Sephardic Jews that had a lower value of 4.8. The gene diversity showed values that ranged between 0.95 and 0.98, and the random match probability was between 5% and 10%.

The eight Y-specific loci showed 71 different haplotypes in 71 unrelated males, therefore all haplotypes were unique.The discrimination capacity was 100% and the haplotype diversity was 1.00.

The data on mtDNA and Y-chromosome were compared between them and with those on autosomical markers previously studied in these populations. With respect to mtDNA, Jewish populations showed a considerable differentiation between them. These results agree with Thomas et al. [3], who indicated that each of the different Jewish communities formed independently around distinct groups of maternal founders and that subsequent gene flow from the host populations was limited on the female side. Autosomal microsatellite data showed no significant differences between all four Jewish populations studied or between them and other circum-Mediterranean populations [4]. Y-chromosome data showed that all the Jewish populations grouped together, and presented a clear

Table 1
Frequencies (%) of the haplotypes observed in the Jewish populations found in at least two individuals

HVRI sequence (sites 15994–16400)	Ashkenazi (20)	Oriental (23)	N. African (33)	Sephardic (31)
CRS	10.0	0	9.1	9.7
224 (C), 234 (T), 311 (C)	15.0	0	0	0
224 (C), 311 (C)	15.0	0	0	0
223 (T), 224 (C), 234 (T), 311 (C)	10.0	0	0	0
129 (A), 223 (T), 264 (T), 270 (T), 311 (C), 319 (A), 362 (C), 391 (A)	10.0	0	0	0
126 (C), 292 (T), 294 (T), 296 (T)	0	17.4	0	0
69 (T), 145 (A), 261 (T), 290 (T)	0	8.7	0	0
284 (G), 362 (C)	0	17.4	0	0
134(T), 189 (A), 223 (T), 278 (T), 311 (C)	0	0	15.2	0
126 (C), 362 (C)	0	0	9.1	0
129 (A), 223 (T), 311 (C), 391 (A)	0	0	6.1	0
298 (C)	0	0	3.0	3.2
356 (C)	0	0	3.0	3.2
86 (C)	0	0	0	6.5
218 (T), 328 (A), 362 (C)	0	0	0	9.7
311 (C), 362 (C)	0	0	0	6.5

Modal haplotypes in each group are underlined. Number of individuals is in brackets.

differentiation with respect to the non-Jewish populations. Therefore, sex-specific differences can been observed in Jewish populations and this fact must be taken into account for a suitable choice of databases in order to weight correctly the value of the evidence of a mtDNA and/or Y profile match.

References

[1] H. Ben-Sasson, A History of the Jewish People, Harvard Univ. Press, Cambridge, 1976.
[2] U. Ritte, E. Neufeld, E.M. Prager, M. Gross, I. Hakim, A. Khatib, B. Bonné-Tamir, Mitochondrial DNA affinities of several Jewish communities, Hum. Biol. 65 (3) (1993) 359–385.
[3] M.G. Thomas, M.E. Weale, A.L. Jones, M. Richards, A. Smith, N. Redhead, A. Torroni, R. Scozzari, F. Gratrix, A. Tarekegn, J.F. Wilson, C. Capelli, N. Bradman, D.B. Goldstein, Founding mothers of Jewish communities: geographically separated Jewish groups were independently founded by very few female ancestors, Am. J. Hum. Genet. 70 (2000) 1411–1420.
[4] A. Picornell, C. Tomàs, G. Jiménez, J.A. Castro, M.M. Ramon, Jewish population genetic data in 20 polymorphic loci, Forensic Sci. Int. 125 (2002) 52–58.