

mtDNA Analysis in a Linguistic Isolate with Different Sampling Strategies

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Introduction

Sampling strategy is a crucial issue in population genetics and anthropological studies, and it is especially important in isolated populations (Destro-Bisol *et al.*, 2008). The sampling choice is related to the research question and the type of markers used. In this study we analyzed the linguistic isolate of Carloforte (Sardinia, Italy) through mtDNA HVRI sequences to verify if different sampling strategies could provide different results and lead to alternative interpretation of the data. Carloforte is the only village located on the small Island of San Pietro; it's an alloglot community founded in 1738 by Ligurian refugees coming from the Tunisian island of Tabarka. Even today the spoken language is the ancient dialect of Pegli (Liguria). Biodemographic data attest a high rate of endogamy and consanguinity, that has slowly been decreasing since 1990 (Vona *et al.*, 1996).

Materials and Methods

Two different samplings were carried out from Carloforte population: the first sampling (N=49) was carried out through grandparents criterion: individuals selected for the study were unrelated and born and resident in Carloforte for at least 3 generations (G). The second sampling (N=50) was based on founders surnames (FS) and the selected participants were proved to be descendants of the village founders, and to have no ancestors in common, at least up to the grandparental generation. DNA was extracted through peripheral blood, and HVRI segments were sequenced. The haplogroups were determined with Phylotree (<http://www.phylotree.org/tree/main.htm>) (van Oven and Kayser, 2009). Since attribution to haplogroup H with Phylotree was sometime hard, the data were confirmed by restriction analysis for the specific polymorphism 7025 Alul (Passarino *et al.*, 1996). Genetic distances were computed using Arlequin program (v. 3.11) and depicted by Neighbour joining tree.

Results

The group selected through FS showed a greater gene diversity, which was confirmed by both network and haplogroup analysis. Among the shared haplogroups, we find clear differences in their frequencies (i.g. H: 33% and 53% and X: 16% and 2% in FS and G sample respectively). Sampling through grandparents criterion showed essentially the same haplogroups found in Sardinia, and with similar frequencies.

The differentiation test between the samples of Carloforte, carried out with Arlequin program, gave a p-value highly significant ($p < 0.0001$). HVRI sequences of the two different Carloforte samplings were compared with the sequences of other Sardinian and Mediterranean populations (<http://www.phylotree.org/tree/main.htm>; Varesi *et al.*, 2000; Vona *et al.*, 2001; Falchi *et al.*, 2006). Carloforte FS sampling is located very far from Italian and other Sardinian populations, in a separate branch, whereas Carloforte grandparents criterion sampling clustered with Italy and it is quite close to other Sardinian communities.

Discussion

Results showed that different sampling strategies can lead to contrasting results. Our results do not imply that one strategy should always be preferred over the other, both strategies being perfectly adequate. We have found a greater variability in the sampling through founder surnames, compared to the sampling through grandparents criterion. Haplogroup analysis and genetic tree analysis also showed significant differences between the two samplings. The contrasting results may simply reflect a different period in the history of the population. Sampling through FS gives indication on the ancient history of the population, while sampling through grandparents criterion is influenced by recent gene flow and therefore provides information on the more recent history, including migration events. The data presented in this paper suggest that Carloforte population changed the gene pool during its history, due to gene flow from the neighbouring Sardinian populations, as witnessed by the increase in mixed marriages in the last two centuries.

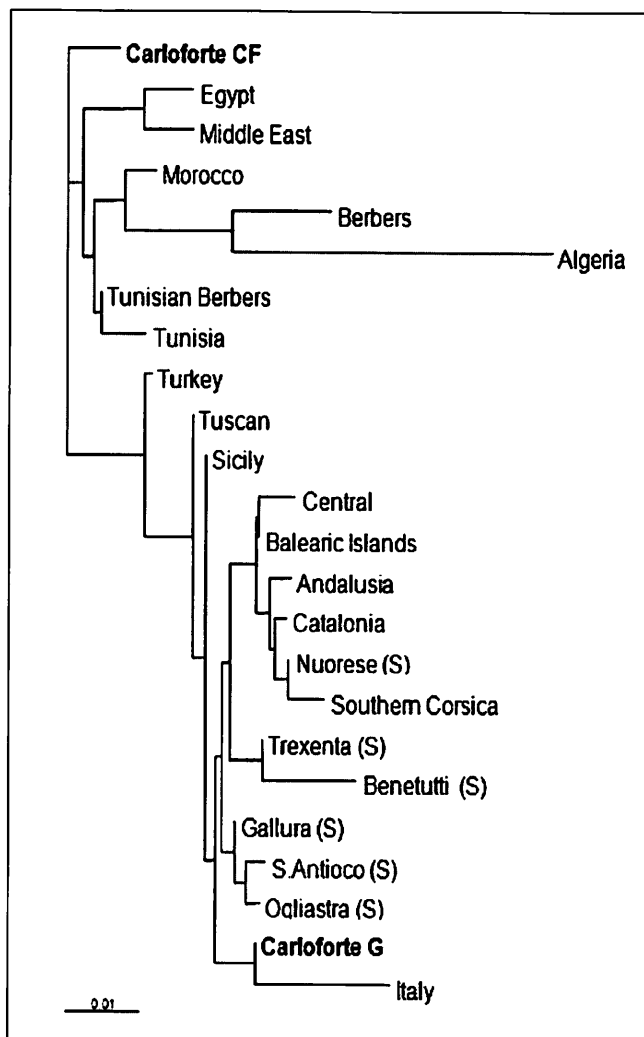


Fig.1. Neighbor joining tree built with the two Carloforte samples, Sardinian (S) and Mediterranean populations.

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