International Congress Series 1288 (2006) 225–227

www.ics-elsevier.com

Y-chromosome haplotypes and male isonymy: Genetic and genealogical study in a small town of Tuscany (Buti, Italy)

Silvano Presciuttini [a,*], Chiara Toni [b], Donato Epiro [a], Isabella Spinetti [b], Fabio Marroni [a], Anna Rocchi [b], Ranieri Domenici [b]

[a] *Center of Statistical Genetics, University of Pisa, Italy*
[b] *Unit of Legal Medicine, School of Medicine, University of Pisa, Italy*

Abstract. We report on 34 subjects from a small Italian town distributed among 13 different surnames. Their genealogies have been reconstructed to the most recent common ancestor, going back in a case to the 15th century, and their haplotypes at 12 Y-chromosome STR has been determined. Six surnames (14 subjects) did not show variation at any locus among isonymous individuals, whereas seven surnames (20 subjects) showed single-step differences at single loci (two in DYS391 and DYS390, the others in DYS385, DYS392 and DYS393); in addition, two subjects of two different surnames showed 8 and 9 locus differences, respectively. © 2005 Published by Elsevier B.V.

Keywords: Y-chromosome haplotype; Genealogy; Surname isonymy; Isolated population

1. Introduction

In societies that use patrilineal family denominations, surnames and Y-chromosome haplotypes (YCH) follow a common pattern of inheritance. Therefore, isonymous individuals are expected to carry the same YCH, and observed inconsistencies can be ascribed to gene mutations, illegitimacies, adoptions, or surname polyphyletic origin. To investigate these issues in a well-defined population, we selected the village of Buti, near Pisa. This small town (about 5000 inhabitants) maintains lively historical traditions; oral

* Corresponding author. Tel.: +39 050 2213797; fax: +39 050 2213524.
E-mail address: sprex@biomed.unipi.it (S. Presciuttini).

0531-5131/ © 2005 Published by Elsevier B.V.
doi:10.1016/j.ics.2005.09.039

anecdotes report high level of reproductive isolation from the neighbouring area. The mayor and the municipality council encouraged participation in the research.

2. Materials and methods

A consent form has been approved by the IRB of the University of Pisa. Volunteers sign it when they donate blood or saliva for the present project at a local health facility. The entire civic records of the municipality have been acquired. In addition, the parochial archives of the main church of the village are almost intact, and were consulted for tracing specific surname genealogies back to the 16th century. YCHs of 12 STR markers were determined by the Y-system Promega® commercial kit. We did not type subjects of first and second degree.

3. Results

To date, 69 males participated in the study; the sampling campaign is still open. The vast majority of the volunteers declared that all four grandparents were born in the village. Here, we report on the subjects whose surname occurred more than once in the sample, and who were unable to specify the degree of relationship with their isonymous fellow citizens. This subsample included 34 subjects distributed among 13 different surnames. Six surnames (14 subjects) did not show variation of YCH

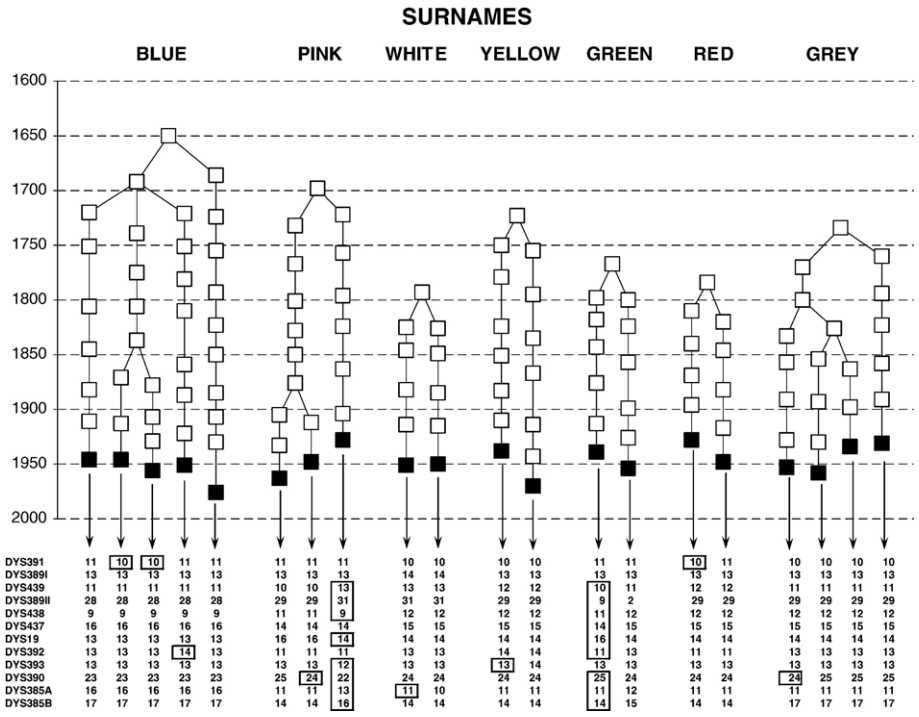

Fig. 1. Y-chromosome haplotypes in 20 individuals of seven different genealogies (surnames). Y-scale: year of birth.

among isonymous individuals, whereas seven surnames (20 subjects) showed one or more locus difference, as follows (see also Fig. 1):

- surname Blue included five subjects; two of whom carried an allele of DYS391 differing of one repeat unit from the allele of the other three, whereas another subject carried an allele of DYS392 differing of one unit from the other four; their genealogy has been reconstructed back to a founder individual (the most recent common ancestor, MRCA) who moved into the village at the end of the 16th century, the total number of meioses separating the MRCA from these five present-day descendants was 37;
- surname Pink included three subjects, two of whom were identical but a single-step difference at locus DYS390, the other totalling 14 and 15 step differences with the first two at seven loci;
- surnames White, Red and Yellow included two subjects each, and showed a single-step difference for the loci DYS390, DYS391, and DYS385, respectively;
- surname Green included two subjects, differing for a total of 11 mutational steps in nine loci. Interestingly, one of these haplotypes was identical to a haplotype observed in Pink.
- Surname Grey included four subjects, one carrying a single-step mutation at locus DYS390.

4. Conclusions

Among 13 different surnames including at least a pair of isonymous individuals (34 total subjects), we were able to identify seven single-step mutations (two in DYS391 and DYS390, the others in DYS385, DYS392 and DYS393) and two cases of historical illegitimacies or adoptions. Our approach shall allow us to estimate mutation rates at STR loci with high accuracy.