



Constituting a Y chromosome short tandem repeats loci database in Sicily

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Abstract. Many Y chromosome short tandem repeats (STRs) have been studied and characterized over the past years. A few Y-STRs multiplex kits have been placed on the market for forensic and population study purposes. The aim of our work was to analyse a sample of Sicilian male individuals to evaluate the allelic frequency and, thus, the possibility to implement a genetic database. An 11 loci Y-STR Typing kit was used to yield haplotype profiles from male DNA; amplification products were detected on ABI PRISM 310 Genetic Analyzer and examined by Genemapper v 3.2 (Applied Biosystems). The population sample subjected to the screening resulted to be very different with regards to certain loci whereas for other loci the allelic profile was less variable. Despite a minor discrimination power within the entire population, Y-STRs represent a valid tool to simplify male/female DNA mixture interpretation which is a major challenge when biological traces are found in case of sexual assault. © 2006 Published by Elsevier B.V.

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1. Introduction

Short tandem repeats (STRs) polymorphisms are mainly used in the forensic field to carry out paternity testing and personal identification. Due to the rising number of crimes

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Table 1
Allelic frequencies of genetic markers present on Y chromosome (%)

Allele	DYS391	DYS389I	DYS439	DYS389II	DYS438	DYS437	DYS19	DYS392	DYS393	DYS390	DYS385
7					1.4						
8					4.1						
9	6.8				39.7						
10	66.2		5.5		34.2			2.9			4.4
11	24.3		37.0		5.5			60.4			16.8
12	2.7	20.2	39.6		13.7			4.4	36.5		4.4
13		62.2	15.1		1.4		9.6	27.9	40.5		15.3
14		17.6	1.4			43.8	42.5	4.4	16.2		19.0
15			1.4			31.5	41.0		5.4		11.7
16						23.3	5.5		1.4		12.4
17						1.4	1.4				9.5
18											3.6
19											2.9
21										1.4	
22										12.3	
23										35.6	
24										43.8	
25										5.5	
26										1.4	
28				10.8							
29				44.6							
30				29.7							
31				9.5							
32				5.4							

committed all over the world, the concern about the creation of different genetic databases has greatly increased in the last years [1]; as a forensic service we have decided to study allelic Y-STR distribution among Sicilian population (Sicily is an island in the South of Italy). The outcomes of our analyses have been deeply studied for different purposes: firstly to compare data obtained from neighbouring geographical areas [2] in order to understand whether Y-STR distribution can vary within the island. Secondly to evaluate how Y-STR allelic distribution varies in respect with the rest of European and Italian population.

2. Materials and methods

2.1. Sample preparation and PCR amplification

EDTA treated blood was collected from 300 unrelated Sicilian. DNA was extracted using the chelex extraction protocol (Biorad). Amplification by PCR of the STR loci was performed using PowerPlex[®] Y System kit (Promega), according to the manufacturer's recommendations [3].

2.2. STR typing

The detection of alleles was performed using AB Prism 310 Sequence Analyzer followed by GeneScan[®] Analysis and Genotyper[®] 3.7 analysis softwares (Applied Biosystems). The length of the amplified DNA fragments was determined based on internal lane standard ILS-600 (Promega).

3. Results and discussion

Our sample consisted of 300 unrelated male individuals. Table 1 shows the observed allelic frequencies distribution in our Sicilian male population sample.

Interesting variations of allele frequency have been observed in some Y-STR loci as DYS438 and DYS385, which turned out to be the most polymorphic loci even among people coming from close geographical areas of the island. In fact, despite their geographical proximity, the different historical origins of these populations were expected to yield meaningful values in the allelic frequencies. This could be a valuable data in determining the area of origin of criminals born and/or residing in Sicily. For the above-mentioned reasons, in the next future we aim to apply Y-STR genetic investigations to an increasing number of genetic markers which may enforce discriminating capabilities.

References

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