Y-Filer Plus® genetic characterization of Caucasian individuals from South Portugal

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Introduction

- Due to their paternal inheritance Y-STRs offers particular perspectives for identification and kinship analysis and are also a precious tool in sexual assault cases with relatively high amount of female DNA and also in mixtures from multiple male donors [1].
- Nonetheless their value, there are some limitations to their use in forensic investigations since their ability to discriminate between individuals is considerably lower than that of the autossomal STRs set, mainly in cases with close or distant patrilineal relatives.
- One of the most recently developed Y-STR kit, Y-Filer Plus® (Life Technologies, Foster city, USA), allows forensic geneticists to study 27 Y-chromosomal loci. All the 16 markers included in the Y-Filer® kit (Life Technologies, Foster city, USA), plus 9 additional markers: DYS576, DYS627, DYS460, DYS518, DYS570, DYS449, DYS481, DYF387S1 and DYS533, six of which (DYS576, DYS627, DYS518, DYS570, DYS449 and DYF387S1) are characterized as "rapidly mutating", and can differentiate between unrelated individuals and possibly between male relatives (Fig. 1) [2].
- In this work it is our goal to characterize south Portugal (Fig. 2) Caucasian individuals, living in South Portugal.

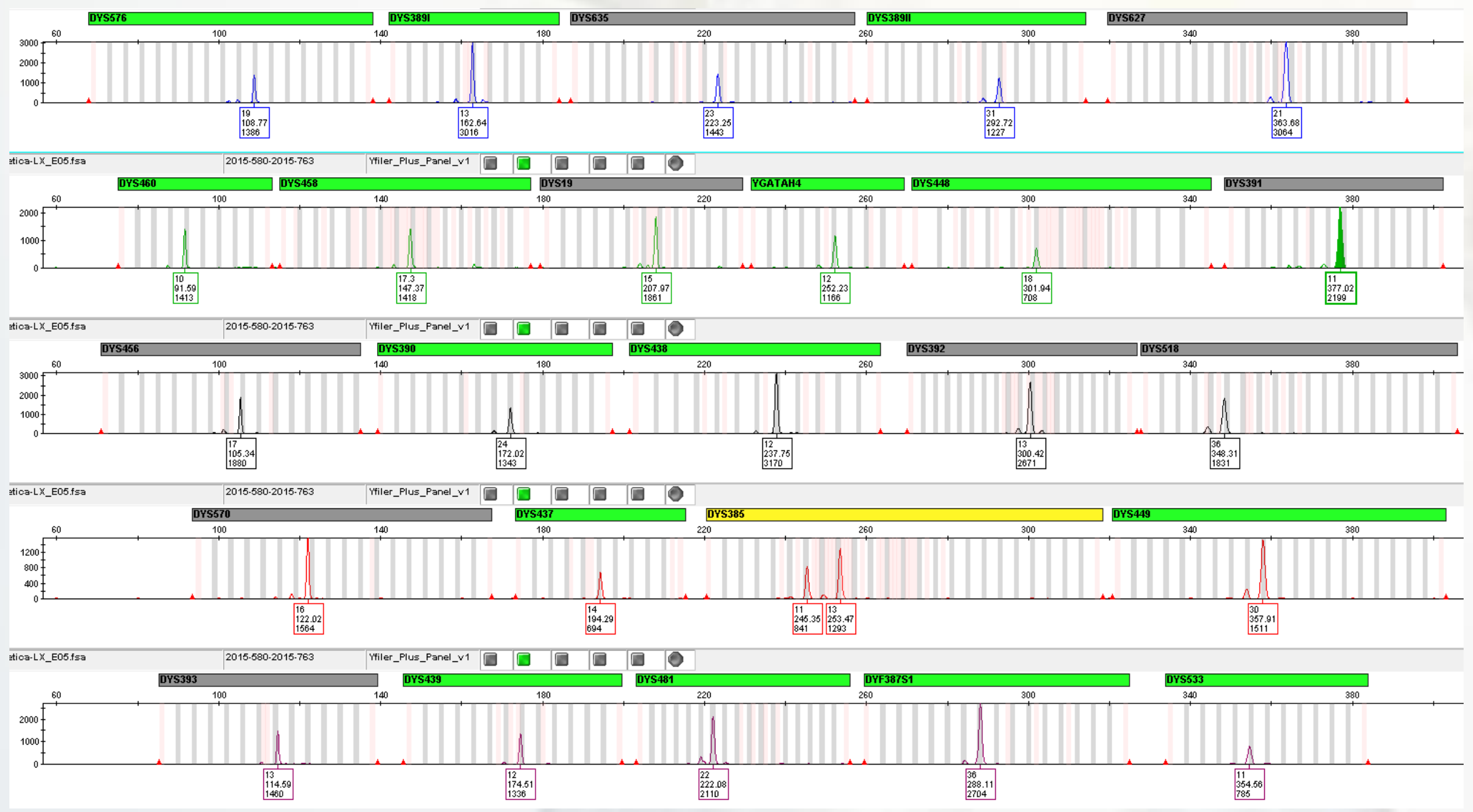

Fig. 1- male Y-STR profile, obtained with Y-Filer Plus® kit (Applied Biosystems)

Fig.2- Map of South Portugal .

Material and Methods

Sample collection and DNA extraction

- Casework routine blood samples from 150 caucasian healthy male individuals. Genomic DNA was extracted using the chelex®100 method as described by Walsh et al.

DNA typing

- The 27 Y-STR markers included in AmpF/STR Yfiler®Plus kit (Life Technologies) were co-amplified using an Applied Biosystems GeneAmp® PCR Veriti® Thermal Cycler in a 6.25 μL volume reaction.

- Amplified products were separated by capillary electrophoresis using an ABI 3130 xl Genetic analyser (Applied Biosystems) using GeneScan ™ 600 Liz v. 2.0 as internal size standard. Sample data were analysed together with an allelic ladder and positive and negative controls using Genemapper® ID-X v 3.2.

Data analysis

Allelic frequencies were estimated with Arlequin v. 3.5. Gene and Haplotype diversities were estimated according to Nei formula. Discrimination capacity was also calculated by dividing the number of different haplotypes by the total number of individuals in the sample. Fraction of unique haplotypes was determined as the percent proportion of unique haplotypes.

Results

- Allele frequencies and gene diversity values for each locus were determined. Gene Diversity (GD) is presented in table 1.
- DYS627, DYS518 and DYS449 showed the highest GD (0.8424, 0.8349 and 0.8303). DYS393, DYS391 and DYS389I showed the lowest GD (0.4628, 0.5506 and 0.5492). Multicopy DYF387S1 reached a GD value of 0.9016.
- Intermediate alleles 16.2, 17.2 and 18.2 were detected in DYS458 *locus*. DYS448 showed copy number variants 19,22 and 18,20.
- The median number of alleles observed per single locus was six, ranging from 4 (DYS389II) to 12 (DYS449).
- A total of 150 unique haplotypes were detected with this 27 Y-STR typing system.
- Haplotype frequency was 0.00676 for each haplotype.
- Haplotype Diversity was 0.999993.
- Discrimination capacity in the sample was 100 %.

Table I- Gene diversity (GD) values for each locus

Locus	**DYS576**	**DYS389I**	**DYS635**	**DYS389II**	**DYS627**	**DYS460**	**DYS458**	**DYS19**
(GD)	0.8011	0.5492	0.6697	0.7192	0.8424	0.5708	0.7821	0.6446
Locus	**YGATAH4**	**DYS448**	**DYS391**	**DYS456**	**DYS390**	**DYS438**	**DYS392**	**DYS518**
(GD)	0.6104	0.7149	0.5506	0.6571	0.6937	0.6391	0.6217	0.8349
Locus	**DYS570**	**DYS437**	**DYS449**	**DYS393**	**DYS439**	**DYS481**	**DYS533**	
(GD)	0.7813	0.6076	0.8303	0.4628	0.6675	0.7306	0.5933	

Discussion and Conclusions

Quality of Y-STR analysis for forensic purposes has been continually improved over the past decades.
With an increase in *loci* number included in this kit, Yfiler Plus provide a higher power of forensic discrimination . The higher number of unique haplotypes, 100% in this sample, results from the large number of markers and the inclusion of rapidly mutating Y-STRS.
The recently introduced Y-Filer Plus® system provides innovative discriminatory power for forensic application.

References

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