STUDY OF GENETIC MARKERS WITH MEDICAL-LEGAL AND FORENSIC INTEREST IN LISBON'S POPULATION (PRELIMINARY RESULTS)

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INTRODUCTION

The population in this study is composed of individuals who were born and live in Lisbon and whose parents were also born in Lisbon.

Due to the lacking of population studies regarding the following new STRs (Short Tandem Repeats): D2S1360, D3S1744, D4S2366, D5S2500, D6S474, D7S1517, D8S1132, D10S2325, D12S391, D21S2055, it was necessary to evaluate their frequencies in Lisbon's population.

Often, in complex cases, in order to confirm previously obtained results, the Investigator HDplex Kit (Qiagen) amplifies a set of STR markers, most of which are not featured in commonly used marker standards. This enables the reliable differentiation of forensic samples providing a heightened discriminatory power in these demanding cases.

The aim of this study is to analyze the frequencies of these additional STRs among Lisbon's population.

MATERIAL AND METHODS

- Blood samples were randomly selected and analyzed from 176 unrelated and healthy individuals, from both genders, without restrictions regarding their ages, from ongoing paternity investigations at the INMCLF, I.P..
- The Chelex 100® method was used to extract the DNA from the selected samples, and then amplified using a 9700 Perkin Elmer (Applied Biosystems) thermocycler following the manufacturer instructions.[1]
- Amplified products were detected using an ABI PRISM® 3130 xl Genetic Analyzer (Applied Biosystems) and fragments were then typed with GeneMapper® ID.
- Allele frequencies and statistical parameters were estimated with Arlequin 3.5.2.2 software and PowerStats V12.[2]

RESULTS

The results of the forensic parameters are shown in table 1.

Table 1: Statistical parameters of forensic interest for 12 autossomal STRs from Great Lisbon's population.

	D7S1517	D3S1744	D12S391	D2S1360	D6S474	D4S2366	D8S1132	D5S2500	D18S51	D21S2055	D10S2325	SE33
Ho	0.9023	0.8621	0.9023	0.88506	0.78736	0.83333	0.82184	0.76437	0.82184	0.86782	0.80460	0.9195
He	**0.86914**	**0.81263**	**0.88835**	**0.87103**	**0.78512**	**0.82200**	**0.85891**	**0.80968**	**0.88221**	**0.88451**	**0.86689**	**0.9513**
P	0.15821	0.00270	0.60823	0.35409	0.32951	0.69968	0.76445	0.32032	0.15773	0.08501	0.30313	0.3555
MP	**0.039**	**0.067**	**0.035**	**0.036**	**0.088**	**0.062**	**0.041**	**0.058**	**0.031**	**0.047**	**0.036**	**0.056**
PD	0.961	0.933	0.965	0.964	0.912	0.938	0.959	0.942	0.969	0.953	0.964	0.944
PIC	**0.85**	**0.79**	**0.85**	**0.85**	**0.75**	**0.79**	**0.84**	**0.78**	**0.86**	**0.84**	**0.85**	**0.86**
PE	0.800	0.719	0.768	0.764	0.576	0.660	0.638	0.535	0.632	0.721	0.608	0.523
TPI	**5.12**	**3.63**	**4.41**	**4.33**	**2.35**	**2.98**	**2.79**	**2.12**	**2.74**	**3.65**	**2.56**	**2.06**
Hom	0.098	0.138	0.113	0.116	0.213	0.168	0.179	0.236	0.182	0.137	0.195	0.242
Het	**0.902**	**0.862**	**0.887**	**0.884**	**0.787**	**0.832**	**0.821**	**0.764**	**0.818**	**0.863**	**0.805**	**0.758**

Ho: observed heterozigosity. **He:** expected heterozigosity. ***P:*** Hardy-Weinberg. **MP:** matching probability. **PD:** power of discrimination. **PIC:** polymorphic information content . **PE:** power of exclusion. **TPI:** typical paternity index**. Hom:** homozygotes. **Het:** heterozygotes.

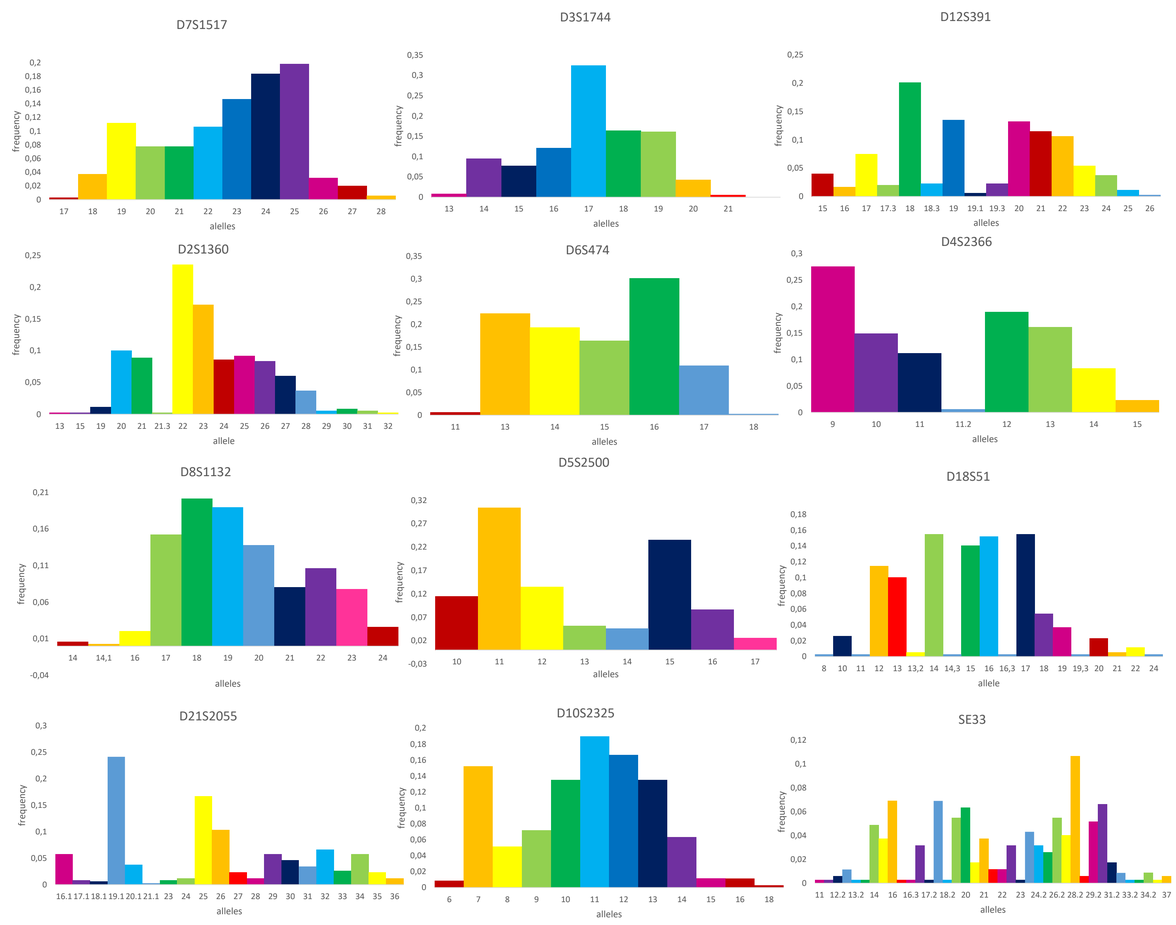

Figure 1: Allele frequencies of the studied genetic markers.

All the markers showed a high degree of genetic polymorphism, with H_e values ranging from 0,785 (D6S474) to 0,951 (SE33). Based on heterozygosity and polymorphic information, the SE33 locus is the most informative among the 12 STR loci. All the PD values were higher than 0,912, and PIC values were higher than 0,750.

CONCLUSIONS

The obtained results for forensic statistical parameters such as observed and expected heterozygosity, polymorphism information content, power of discrimination, based on single allele frequencies reveal that the Investigator HDplex Kit (Qiagen) is highly informative and can represent an important tool for genetic identification purposes for forensic investigations ongoing at the INMLCF, specially in paternity cases, related to individuals from the great Lisbon area.

Bibliography

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