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STR and HLA analysis in paternity testing

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Abstract. During the 3-year period, 111 cases of paternity disputes were processed in our laboratory. The analysis was performed on nine STR (TH01, VWA31, FES/FPS, F13A01, SE33, D1S1656, D12S391, D18S535, and D22S683) loci and one VNTR (D1S80) locus. Out of 111 cases, 22 were exclusions. In all cases, exclusion was confirmed on at least five loci. STR loci that were informative in majority of cases were SE33 and D12S391. The least informative loci were FES/FPS and F13A01. In 21 cases exclusions were also confirmed by HLA class I, while only in 17 cases HLA class II alleles excluded the alleged father. © 2003 Elsevier B.V. All rights reserved.

Keywords: Paternity testing; HLA; STR

1. Introduction

HLA system is the most polymorfic genetic system in human genome so far known and for that reason it was used in paternity testing for a long time. However, due to the predominance of certain HLA alleles and linkage disequilibrium, in some cases HLA was not informative enough. During the past decade STR loci became a valuable tool in paternity testing because of their high polymorphism and heterozygosity [1]. The mutation rate of STR loci is higher than that of HLA or some other genetic markers. Combination of these two systems offers higher informativeness and also diminishes the possibility of false exclusion due to mutations.

The aim of this study was to investigate and compare the usefulness of these two systems in cases of paternity dispute.

2. Material and methods

One hundred and eleven triplets (mother, child, and alleged father) were processed in our laboratory during the 3-year period. Nine STR loci (TH01, VWA31, FES/FPS,

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| Locus | Percentage of informativeness |
|---------|-------------------------------|
| TH01 | 62.4 |
| VWA31 | 62.2 |
| FES/FPS | 55.6 |
| F13A01 | 57.9 |
| SE33 | 85.7 |
| D1S80 | 66.3 |
| D1S1656 | 74.3 |
| D12S391 | 82.9 |
| D18S535 | 68.5 |
| D22S683 | 76.2 |

Table 1 The list of informativeness for nine STR loci and one VNTR locus

F13A01, SE33, D1S1656, D12S391, D18S535, D22S683) and one VNTR locus (D1S80) were tested using four different PCR amplification protocols [1–4]. The analysis of the PCR products was performed by electrophoresis on a 6% polyacrilamide gel in an automated laser fluorescence sequencer (ALFexpress, Pharmacia Biotech). Assignment of alleles was done with Allele Locator program. HLA class I (HLA-A, -B) and class II (HLA-DR) antigens were detected by standard Microlymphocytotoxicity test in Terasaki plates using local and commercial sera.

3. Results

As one can see from Table 1, STR loci that were informative in majority cases were SE33 (85.7%) and D12S391 (82.9%). The least informative loci were FES/FPS (55.6%) and F13A01 (57.9%).

Out of 111 cases, 22 (19.8%) were exclusions. In all cases, exclusion was confirmed on at least five STR loci (five triplets). Ten exclusions were confirmed on six loci, four exclusions at seven loci while three exclusions were documented on eight loci (Fig. 1). We did not find any triplet with exclusion on all 10 loci.

The distribution of exclusions for each STR locus is presented in Fig. 2. The highest number of exclusions was observed at SE33 (18 cases) as opposed to TH01 (7 cases). On the other hand, analysis of HLA polymorphism showed exclusions in 21 out of 22 exclusions.

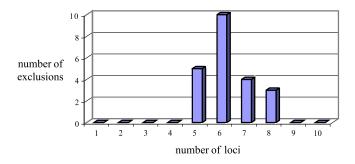


Fig. 1. Distribution of cases according to the number of exclusions.

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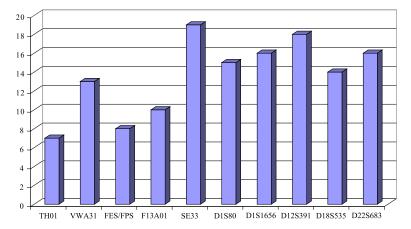


Fig. 2. The distribution of STR loci according to the number of exclusions.

In conclusion, analysis of STR loci combined with HLA polymorphism offers more reliable results for resolving paternity disputes.

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