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GEP proficiency testing program in forensic genetics: 10 years of experience

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Abstract. Since 1992 the GEP-ISFG group has been organizing a proficiency testing program open to all the laboratories in the group. The number of participating laboratories increased continuously from 10 in the first exercise (GEP'93) to 89 registered for the last exercise (GEP'02), 86 (95%) having submitted results. Despite the increasing number of laboratories, results remained quite satisfactory. There is a tendency to concentrate errors in a few laboratories and also an association with the use of home made ladders. mtDNA and statistics are the challenges nowadays, and actions have been implemented in the group to solve the difficulties found in the laboratories for this type of expertise. © 2003 Published by Elsevier B.V.

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The Spanish and Portuguese Working Group of the International Society for Forensic Genetics (GEP-ISFG) (www.gep-isfg.org) comprises forensic genetics laboratories from Spain, Portugal, France and most of the Portuguese and Spanish speaking countries in America. A total of 324 forensic geneticists from 118 laboratories and 19 countries are members of the group. Spain, Portugal, Brazil, Argentina and Colombia are the countries more represented but the group include most of the laboratories performing forensic genetic casework in the area. Almost all the laboratories are involved in paternity testing but only half (58%) of the laboratories are in criminal casework.

Since 1992 the GEP-ISFG working group has been organizing annual collaborative exercises on DNA profiling with the aim of making progress in standardization and discussing technical and statistical problems in DNA profiling. Since 1995 a proficiency testing program coordinated by Quality Control Unit (National Institute of Toxicology Madrid) was carried out simultaneously with the GEP-ISFG collaborative exercises. The number of participating laboratories increased continuously from 10 in the first exercise

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(GEP'93) to 89 registered for the last exercise (GEP'02), 86 (95%) having submitted results.

The samples included in the proficiency testing are decided by the QC office and include a number (usually five to six) of 100 μl bloodstains (including a paternity testing case), a 2 cm hair shaft (from 2000) and forensic samples, including saliva samples (1997), mixed stains (1998, 2000, 2003), seminal stains (2002) and nonhuman samples (2001). Statistical evaluation of the evidence was required in the last trials, including a theoretical paternity testing case with reported frequencies. Only systems with at least three laboratories having consensus are considered in the PT trial and this number will increase up to five laboratories from 2003. In case of discrepancies in one or more system results are studied by a panel of experts and discussed in the GEP meetings.

All the laboratories used autosomal STRs with an increasing number of markers until recently (now the tendency is for a stabilization in the number of markers used and a policy has been implemented to achieve the goal of a reduction in the number of systems). SLPs were used by a decreasing number of laboratories, and hardly a couple of laboratories have submitted results in the last trial. The same occurs for dot-blot based systems (HLADQA1, Polymarker). On the contrary the mtDNA and Y chromosome are increasingly being used by the laboratories (Fig. 1).

Despite the increasing number of laboratories, results remained quite satisfactory. The total number of errors ranked from 0.6% to 2.3% in the different years (average 1.5%). In general, there is a tendency to concentrate the errors in a few laboratories most of them using manual electrophoretic systems and home made allelic ladders. In the last trial, only five laboratories (10%) are responsible of 50% of errors. In addition if only STRs included in commercially available kits are included the error rate dramatically decreases with an average of 0.65%.

Y STRs have a similar error rate than autosomal STRs and the group have made a great effort towards the standardization of new Y STRs [1], including the compilation of population data [2]. The results of mtDNA are quite variable, depending in the difficulties

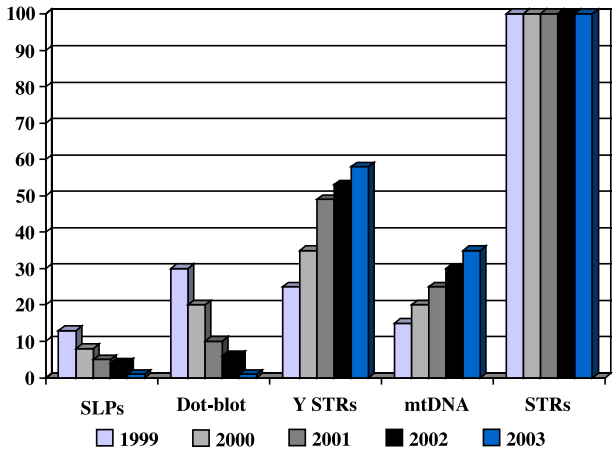

Fig. 1. Percentage of laboratories using the different types of markers.

of the sample. In some case heteroplasmy was detected making more difficult interpretation. The difficulties found in two different years one related with the low/degraded amount of DNA present in the hair shaft samples submitted [3] and other due the variation of heteroplasmy in different hair samples [4] has been recently published.

The group is aware of the need to improve mtDNA results. Similar results were observed in other collaborative exercises or population data compilation. To follow strictly ISFG recommendations regarding contamination, the use of validated software and especially phylogenetical checking of the data are strongly recommended. An important number of statistical errors in statistical programs and calculations were detected in the theoretical paternity testing cases (paper challenges), especially when the difficulties of the case increase. This indicates that a greater effort must be made in this area and the corresponding actions to correct this problem have been implemented, through training courses on basis of statistical evaluation and the use of validated software in various countries of Europe and America.

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