



The proficiency testing program on DNA typing of the Spanish and Portuguese working group of the International Society for Forensic Genetics

Josefina Gómez, Antonio Alonso, Oscar García, Angel Carracedo*,
on behalf of the Spanish and Portuguese ISFG
Working Group (GEP-ISFG)

Institute of Legal Medicine, Faculty of Medicine, 15705 University of Santiago de Compostela, Galicia, Spain

Abstract

The Spanish and Portuguese Working Group (GEP) of the International Society for Forensic Genetics (ISFG) comprises members from 90 forensic genetic laboratories from Spain, Portugal and most of the Portuguese and Spanish speaking countries in America (<http://www.usc.es/gep-isfh>). The activities of the group since 1992 are described, including collaborative exercises and proficiency testing programs, as well as other activities including the progress made in accreditation.

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The Spanish and Portuguese Working Group (GEP) of the International Society for Forensic Genetics (ISFG) comprises forensic genetic laboratories from Spain, Portugal and most of the Portuguese and Spanish speaking countries in America (<http://www.usc.es/gep-isfh>). A total of 242 experts from 90 laboratories from 15 Iberoamerican countries are members of the group; Spain, Colombia, Argentina, Portugal and Brasil being the countries with a highest number of participants and labs.

There is a variety of labs included in the group from university labs, governmental labs from the Ministry of Justice and Interior to private labs. Some labs are devoted to criminal casework and others to paternity testing only, but the majority perform both.

* Corresponding author. Tel.: +34-981-582327; fax: +34-981-580336.

E-mail address: apimlang@usc.es (A. Carracedo).

The great majority of labs (close to 100%) performing forensic genetic analysis in Iberoamerica are members of the group.

As with other working groups of the ISFG (<http://www.isfg.org>), since 1992, the GEP-ISFG has been organizing annual collaborative exercises on DNA profiling with the aim of making progress on standardization and discussing technical and statistical problems in DNA analysis [1,2].

The first collaborative exercise was carried out in 1992; 10 labs participating in the exercise and 2 SLPs, D1S80 and HLA DQA1 were included in the exercise. The number of participants and the activities of the group increased considerably in the following years and one of the consequences was the creation of a proficiency testing programme in Spain and Portugal in 1995, which was carried out simultaneously with the GEP collaborative exercises. This PT scheme is coordinated by the Quality Control Office of the National Institute of Toxicology (Spanish Ministry of Justice). The QC office distributes samples (a paternity case is always included) and statistical cases with detailed information on the exercise, in addition to a questionnaire. The labs can submit the results of the proficiency testing trial or simply as a collaborative exercise. In the first case, the QC office will certificate the results obtained in the trial and the action undertaken by the labs to correct any problem detected.

The results are discussed in the annual meeting of the GEP group. The next one is going to be held in Cordoba (Argentina) in October.

mtDNA was included in the PT trial from 1996. In this exercise, a criminal case (with saliva stains) was included in addition to the paternity case.

Y chromosome STRs were added in 1997 and the criminal case included a mixed stain. The number of labs in this exercise increased to 28 and the number of markers used to 52.

In the GEP '98 trial, a theoretical paternity case was included and the frequencies of the alleles given for statistical evaluation. A high number of theoretical mistakes in participants were detected and some errors in some statistical programs were also found.

The results were considerably better in 1999, but when the difficulties of statistical analysis increased in the 2000 exercise, new problems were detected. The conformity of results achieved in the paternity case and in the criminal case is a remarkable finding and a good indication of progress in statistical standardization. The theoretical paternity case included in the trials proved to be a good exercise to detect statistical problems and software errors. This indicates that a greater effort must be made in this area and considerably progress in statistical standardization has been achieved in the group, thanks to this exercise.

In the GEP '99 trial, a maternity testing case was also included and the number of participants and markers considerably increased.

The number of participating laboratories increased from 10 in the first exercise (GEP '93) to 77 in the last exercise (GEP '00) and it has been continuously increasing each year.

Some other labs (out of the group) are using the GEP-PT program as an external PT trial and the number of labs from America is now higher than the number of European labs.

Concerning the number of markers used by the different labs, the number of STRs was continuously increasing until 1999, but in the last exercise, it has started to decrease. The

number of minisatellites and dot-blot systems has been continuously decreasing from 1995. Now, it has more or less stabilized as a small group of these type of marker. By contrast, the number of Y STRs used by the different labs is increasing.

The same tendencies are observed if we consider the markers used by at least three laboratories. The QC office only certificate results reported by at least three labs. In this case, the number of STRs used decreases considerably, but the same global tendencies are observed for all the markers.

STRs are used now by all the labs including those working only in paternity testing. There is a clear tendency towards an increasing use of Y STRs and a slight tendency towards the introduction of mtDNA in more labs. On the other hand, dot-blot systems and SLPs are decreasingly being used and now, they have been used only by a few labs.

The error rate was in the range of 0.5–0.8 in the first 5 trials, with the introduction of many new labs with little experience, the percentage of errors increased up to 2%. However, there has been a reduction in the number of errors in the last trial and there is a clear tendency towards a concentration of errors in the same labs and with just two labs excluded, the number of errors remains around 0.75%, which we consider to be a good standard from a technical point of view.

Concerning the type of error, most are technical and they are usually related to a lack of automation or the use of home-made ladders. Some of the errors are transcriptional and others are related to problems of nomenclature. This is an important problem for the Y STRs, specially for the new STRs and the corresponding actions should be undertaken by the scientific community to solve the problem of nomenclature.

Despite the difficulties of mtDNA analysis, and the fact that heteroplasmies were observed in the last exercise, the results were also very satisfactory. There was a significant increase in the number of participants and the detailed analysis of results led to very interesting conclusions in the statistical evaluation of the evidence in cases of mtDNA matches.

In addition to the proficiency testing and the collaborative exercises, GEP also has very active working groups. This includes the WG on accreditation which has produced standards for accreditation for forensic genetics, which have been accepted by all the labs. Other working groups are those concerned with sample collection for DNA testing, with nuclear and mtDNA polymorphism databases, with Y STRs, and with evaluation and training which also includes a working group on standardization of statistical evaluation of the evidence. There is also a working group on ethical problems in forensic genetics and the standardization of criminal databases.

These collaborative exercises together with the Quality Control Programme have proven to be extremely valuable and clearly improve the quality of the medico-legal expertise in forensic genetics in Iberoamerican countries.

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