THE ITALIAN EXTERNAL QUALITY ASSESSMENT IN GENETIC TESTING: DEVELOPMENT OF A WEB BASED SYSTEM

Vincenzo Falbo, Giovanna Floridia, Marco Salvatore, Manuela Marra, Fabrizio Tosto, Federica Censi, Domenica Taruscio
Centro Nazionale Malattie Rare, Istituto Superiore di Sanità, Roma

Introduction

The Italian External Quality Control (EQC) started in 2001 at the Istituto Superiore di Sanità (ISS, the National Institute of Health in Italy). The major goal of this activity is the improvement of the performance in genetic tests in the clinical practice by elaboration of recommendation and guidelines, standardization of methods and diffusion of technical information. Participation is free of charge, voluntary and limited to Italian public laboratories.

The EQC programs covers seven schemes four of them in molecular genetics (Cystic Fibrosis - CFTR gene, Beta-Thalassemia - Hbb gene, Fragile-X syndrome - FMR1 gene, the Adenomatous Polyposis Coli - APC gene) and three in cytogenetic (prenatal, postnatal and oncological diagnosis).

Eightytwo public laboratories distributed on the National territory have been enrolled. Five trials have been performed and concluded. Results showed that there has been an improvement in the use and in the interpretation of molecular genetic tests. The average genotyping error rate observed over the five years was 0.6%, 0.3%, 5% and 3.7% in the Cystic Fibrosis, Beta-Thalassemia, Fragile-X syndrome and Adenomatous Polyposis Coli scheme respectively; the percentage of complete reports in cytogenetics increased over the period. However, lack of information or inadequacy in reporting are still observed. On the other hand, as has been indicated in other international surveys for quality assessment, it will be only after several years of testing experience and participation in quality assessment schemes that a significant reduction in laboratory errors will be possible.

Methods

On the basis of the experience acquired until now and in order to harmonize the activity of our schemes with existing European ones, we have developed a web-based system. The web based system is structured into three sections: i) a specific section is restricted to the ISS as scheme organizer; ii) a second section is restricted (through password) to each participating laboratory in order to load, by web, raw data and reports, consulting the manual available on the website; iii) a third section is restricted (through password) to assessors, (decided by Italian Society of Human Genetic for each scheme), that will use this area in order to perform on line the evaluation of the raw data, from they computer; the final evaluation will be performed at the ISS. The web based system allows: a) to handle about 90% less of paper; b) to manage easily materials; c) to archives performance during the years.

Participation in EQA Schemes will be open to all public laboratories enrolled during the five trials and recorded in our database. New laboratories need to be registered contacting by e-mail: testgenetici@iss.it.

Participants are informed in advance of the date of EQA opening either in cytogenetics and molecular genetics. For any problem it is the participant's responsibility to inform the scheme organiser.

Cytogenetics EQA scheme is based on a retrospective format; laboratories send to ISS images and reports of two cases analysed.

A regards molecular genetics EQA, laboratories are asked to treat the samples using current methods and to report the results (raw data and full interpretative reports), via their own account, in the website, in anonymous, and identified solely by their ID. Technical data related to samples and clinical indication must be downloaded from the website The Laboratory that does not provide returns on or before the given deadline will be deemed not evaluated.

At the end of the evaluation, participants will download their performance directly from the website. Participants who wish to present to scheme organiser claim related to their performance evaluation can do so using the address present in the website.

Results

The web based system has been used for the sixth trial. A total of 96 Public laboratories have been enrolled by website through an account, with an increase of 65.3% compared to the first trial. In particular: 35 laboratories out of 96 were registered for molecular genetics, 30 for cytogenetics, and 31 for cytogenetics and molecular genetics. The number of respondents was: 46/49 (94%) for Cystic Fibrosis; 22/23 (96%) for Beta-Thalassemia; 18/21 (86%) for Fragile-X syndrome; 6/6 (100%) for Adenomatous Polyposis Coli; 37/43 (86%) for prenatal diagnosis; 47/54 (87%) for postnatal diagnosis; 28/33 (85%) for oncological cytogenetics.

To better identify the performance of laboratories a marking system has been developed in molecular genetics and in cytogenetics in cooperation with Dr. R. Elles, Dr. S.Patton (European Molecular Genetics Quality Network) and with Dr. Ros Hastings Dr. Rod Howell (Cytogenetic European External Qualità Assessment, EUROGENTEST), on the basis of their long experience in EQA.

At the moment, assessors are evaluating raw data.

In a second time, assessors will attend a meeting at the Italian National Institute of Health to discuss and to perform the final assessment.

Conclusions

The increase of the number of participants (19 responders more than the fifth trial) suggests that a web based system is more easily approachable for laboratories. The efficiency of the proposed marking system will be evaluated in a few years time.

Papers published within the project

Falbo V, Floridia G, Salvatore M, Marra M, Tosto F, Censi F, Taruscio D. The Italian External Quality Assessment (EQA) in genetic tests: the VI EQA scheme. In: Taruscio D, Salvatore M (Ed.). *Workshop. Projects on rare diseases funded within the bilateral agreement Italy (Istituto Superiore di*

Sanità) and USA (NIH, Office for Rare Diseases) on joint research and development of public health actions. Istituto Superiore di Sanità. Rome, October 29-31, 2008. Abstract book. Roma: Istituto Superiore di Sanità; 2008. (ISTISAN Congressi 08/C10). p. 54.

Falbo V, Floridia G, Salvatore M, Tosto F, Censi F, Taruscio D. The Italian external quality assessment in genetic tests: development of a web-based system. In: Taruscio D, Salvatore M (Ed.). *Workshop. Rare Diseases and Orphan Drugs. Rome, November 7-8, 2007. Abstract book.* Roma: Istituto Superiore di Sanità; 2007. (ISTISAN Congressi 07/C8). p. 42.

Acknowledgments

We thank for their scientific contribution Prof. G. Guanti, Dr. Pasini, Dr. P. Radice, Prof. MC Rosateli, Dr. A. Ravani, Dr. M. Grasso, Prof. MA. Melis, Dr. S. Russo, Prof. AM. Baffico, Dr. C. Bombieri, Dr. E. Pelo, Dr. D.Giardino, Dr. E. Lenzini, Dr. A. Novelli, Dr. G. Piombo, Prof. G. Calabrese Prof. C. Mecucci, and Prof. M. Mancini.