Capacity building of health professionals on omics sciences: evaluation of the effectiveness of a distance learning training course for Italian physicians.

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Abstract

**Background.** The rapid adoption of personalized medicine approaches in healthcare requires professionals to be able to manage the “omics revolution”. In this context, the genetics/genomics literacy of healthcare professionals should be in line with the continuous advances in this field, in order to implement its potential implications for the diagnosis, control and treatment of diseases. The present study investigates the effectiveness of a distance learning course on genetics and genomics targeted at medical doctors.

**Methods.** In the context of a project funded by the Italian Ministry of Health in 2017, we had set up a distance learning course, entitled *Genetics and Genomics practice*. The contents of the course were identified from a core curriculum in genetics directed mainly to general practitioners, for physicians and healthcare professionals, previously published. The course focused on genetic/genomic testing in clinical practice, pharmacogenomics and oncogenomic and it was developed according to the main models of andragogical training (Problem-based Learning and Case-based Learning). We used a pre-test versus post-test study design to assess knowledge improvement on a set of 10 Multiple Choice Questions (MCQs). We analysed the proportion of correct answers for each question pre and post-test, as well as the mean score difference stratified by gender, age, professional status and medical discipline. Moreover, the same test was submitted to the participants eight months after the conclusion of the course, in order to assess the retained knowledge.

**Results.** An important number of Italian physicians (N= 1637) completed the course, most of which were primary care physicians (20.8%), public health
professionals (11.5%) and specialist paediatricians (10.6%). We reported an improvement in the proportion of correct answers for all of the 10 MCQs set at the post-test. The overall mean score to the questions significantly increased in the post-test, from 59.46 in the pre-test to 71.42 in the post-test (p-value < 0.0001).

**Conclusions.** Genomic literacy among healthcare professionals is essential to ensure optimal translation to healthcare delivery of research. The results of this course suggest that distance-learning training in genetic/genomic practices represents an effective method to improve physicians’ knowledge in the immediate and mid-term time scale.

**BACKGROUND**

The last two decades were characterized by a "genetic revolution" that has given rise to the “omics sciences era” as a consequence of the spread of high-throughput investigation techniques, capable of generating enormous amounts of data related to the different hierarchical levels of biological complexity (DNA, mRNA, proteins, metabolites, etc. ...) [1].

The rapid spread of this new knowledge requires healthcare professionals to manage the possible application of the omics sciences, ranging from medical research advances to use in screening, diagnosis and prognosis of different pathologies [2, 3].

While several European countries implemented dedicated health policies in this area [4], few countries have integrated Public Health Genomics into the health system offer [5], such as Italy where personalized medicine was included in the National Prevention Plans since 2010 [6, 7].

More recently, the Italian National Plan for innovation of the Health System based
on omics sciences, identified educational efforts towards professionals, citizens and decision makers as a cornerstone for a proper implementation of omics sciences in healthcare [8]. In this context, the Italian Ministry of Health has supported a distance learning course entitled "Genetics and Genomics practice", aimed to train medical professionals and in particular at primary care physicians, in the responsible use of omics technologies [9]. The course included audio-video lectures and interactive clinical cases and it was structured according to the main models of andragogic training (Problem-based Learning and Case-based Learning). The Problem-based Learning (PBL) is a training methodology that stimulates the participants to "learn to learn" by solving real-world problems that reflect their work context [10]. This methodology has been successfully used in other distance learning courses targeted at medical doctors and developed by the same working group [11, 12]. The Case-based Learning (CBL) is a teaching methodology used in medical education field, as an aid in connecting theory to practice [13].

Our study presents the project aimed at developing a distance learning course in genetics and genomics, and to evaluate its effectiveness in terms of knowledge improvement of participants after the course, and eight months after its closing.

METHODS

Elaboration of the scientific contents of the course

The content of the course and the delivery model were identified according to previously two literature reviews: the first systematic and aimed at identifying the core competencies in genetics/genomics for non-genetics healthcare professionals [14], and the second aimed at identifying the most effective educational
interventions to improve knowledge of health professionals in the "omics" context [15]. Subsequently the course topics were validated by a panel of expert geneticists involved as teachers of the course.

The general and specific objectives and the content of the course, including 9 case studies, are reported in Table 1.

Course characteristic, learning methodology and participants

The distance course "Genetics and Genomics practice" was accessible free of charge by the Italian National Institute of Health e-learning platform (EDUISS - https://www.eduiss.it). The Learning Management System (LMS) used was Totara Learn, that offered the technical resources to reproduce the selected methodological approaches (PBL and CBL).

The course was delivered from February 27th, 2017 to February 1st, 2018. The course, open to all physicians potentially involved in the prescription and/or interpretation of genetic tests, was primarily targeted at General Practitioners (GPs) and Family Pediatricians (FPs). The maximum number of subscribers was 2500.

Successful completion of the course included the release of 30 Continuous Medical Education (CME) credits. Participants were expected to spend 30 hours to complete the course and could access the course at any time.

The course was structured in 4 sections:

1. Introductive section: introduction to the course to explain its relevance, general aims and structure; general objectives of the course; participants guide containing all the information needed to attend the course; preliminary self-assessment test to set the initial knowledge and skills competencies (pre-test), consisting in 10 Multiple Choice Questions (MCQs). No minimum score
was required to complete the test.

2. PBL section (1 entire PBL cycle - 7 steps): problem presentation and analysis, specific learning objectives identification, bibliographical references and a list of useful web sites to be consulted, reading materials to deepen the topics of the course, audio-video tutorials by experts and problem’s solution.

3. Case Studies section: exercises on 9 different clinical cases studies (Table 1).

4. Conclusive section: post-test (same MCQs set of the pre-test), final certification test, satisfaction questionnaire. Passing the final test, consisting in 90 MCQs (0 points for wrong answers - 1 point for correct answer) was mandatory to complete the course. The passing mark was set at 75% of the scored questions answered correctly. Three passing attempts were allowed.

The course methodology integrated the PBL [11, 12] and the CBL [13], in order to satisfy the strong clinical orientation of the course.

The entire PBL cycle was set up using platform tools such as feedback, web pages, quizzes. The first steps of the PBL cycle, consisting in problem analysis and learning objectives identification were provided through an interactive tool that allowed the participants’ results tracking.

The case studies were realized through interactive exercises, consisting in clinical case audio-video presentations, animated slides, clinical notes and a final assessment test on the case focal points.

Data collection

When registering for the course in the e-learning platform, the following demographic and professional information about the participants were collected from the system: gender, age, region of residence; CME discipline; professional status (National Health Service - NHS employee, freelancer, private contractors with
NHS, unemployed). A preliminary question on the previous learning experience on genetics was proposed only at the beginning of the course. Afterwards, a pre-test was performed in order to gain insight on the knowledge and skills competencies, at course registration (T0). The same test was repeated after the course only for those who completed the entire course and passed the CME certification test (T1). The post-test was repeated also at eight months after the closure of the course (T2). The test consisted of 10 Multiple Choice Questions (MCQs) related to the different modules of the course. For those completing the test at T2, an additional question on the perceived competence about the capability to meet the patients requests on genetic tests was administered. At the very end of the course, participants which successfully completed the final MCQ test were also required to fill in the satisfaction questionnaire (SQ), consisting in 18 closed questions about the perceived quality of the Learning Methodology, the educational Contents and the E-learning platform functioning.

**Statistical analysis**

We performed a descriptive analysis for demographic and professional information. The results of each question, for the pre-test, post-test and for the follow-up test, were reported as percentages of correct answers. The pre-test and post-test results were compared through the McNemar test. We calculated a score for ten MCQs by assigning 10 points for each correct answer. The average scores of the pre-test and post-test were compared, by t test for paired data for the eligible participants. Data were stratified by gender, age categories, region of residence (North, Center, South and Islands), medical discipline and professional status. The discipline of the course participants was analyzed reporting individually those that reached a number greater than 3% of the total, while those for which a lower number was recorded
were grouped as "other specializations". The results of the follow-up test were analyzed through descriptive analysis, given the reduced number of respondents. Statistical analyzes were performed using the Stata software (StataCorp. 2013. Statistical Software: Release 13. College Station, TX: StataCorp. LP).

RESULTS

Course participants

The participants who completed the course (Completers) were 1637 out of 3054 members enrolled (Figure 1).

Table 2 describes the characteristics of the participants and of the 268 participants who filled in the follow up test (Responders).

Among the Completers, 790 (48.3%) were male and 847 (51.7%) females. The median age was 56 years and the most represented age group was 51-65 years (58.3%). Most of the participants were from the South of Italy (including Islands) (38.5%), followed by North (36.3%) and Center (25.2%). The most represented disciplines were those referring to "primary care" (GPs, FPs, and Continuity of care Physicians), accounting for the 20.8% of participants, followed by "hygiene, epidemiology, health organization" (11%), specialist pediatricians (10.6%), "psychiatry and psychotherapy" (7.2%), "sports medicine" (4.2%), "genetics and laboratory genetics" (3.7%), "occupational medicine" (3.3%) and "gynecology and obstetrics" (3.2%). Regarding the professional status, most of the participants were private health facilities or NHS Employees (55.8%).

The Responders at the follow-up were 268 out of 1637 (16.4%). Among them, 145 (54.1%) were males and 123 (45.9%) females. The mean age was 55 years and the most represented age group was 51-65 years (63.8%). Most of the participants came
from the North of Italy (37.7 %), followed by South (37.3%) and Center (25%). The most represented disciplines were “primary care” (23.5%), “hygiene and public health” (14.9%) and specialist pediatricians (10.8%). As for the professional status, most of the participants were private health facilities/NHS Employees (55.2%).

**Effectiveness of the course**

The preliminary question on a previous attendance to similar training courses indicates that 79.4% of participants had not attended other courses on the same topic. The results of the pre-test versus post-test comparison are reported in Table 3. A significant improvement was registered in 100% of the questions.

Table 4 presents the results of the comparison between average pre-test and post-test scores according to several participants’ characteristics. The average overall pre and post-test scores were 59.5 and 71.4, respectively, with a mean increase of 11.9 (p-value <0.0001). In stratified analysis, a significant improvement in the average scores was recorded for all the categories considered. The stratified analysis by age shows that, with increasing age, the pre-test score was lower, along with a progressive increase in the difference between average pre-test and post-test scores. The stratification by region of residence reports a North-South decreasing gradient both in the pre-test score and in the score increase between post-test and pre-test. The stratified analysis by medical discipline shows the greatest increase in knowledge for “sports Medicine physicians” (score increase 15.3) and for the class of physicians belonging to "primary care" (14.3). These classes had the lower pre-test score (53.1 “sports Medicine physicians” and 57.0 “primary care” physicians). Within the "primary care" class, GPs had the lowest pre-test score (56.3) and achieved the greatest increase (score increase 15.3) (data not shown). With regard to professional status, the highest increase in scores was
recorded for the private contractor with the NHS (14.4), that were those who had the lowest pre-test score (57.9).

Table 5 and Table 6 report the results of the Responders at follow up (T2) test. Regarding the knowledge, with the exception of the question 4 and 5, improvement was recorded when comparing the correct answers to the post test (T1) and to the follow up questionnaire (T2) with respect to the pre-test (T0) (Table 5).

Table 6 reports the results of the Responders to the additional question on the perceived competence about the capability to meet the patients requests on genetic tests. Among the 258 Responders the perceived competence has overall improved: those who felt less competent passed from 45.2% at the pre-test to 41% and 28.2% at post-test and follow-up respectively; and there was an increase in the number of Responders who felt more capable of providing information, from 18.2% in the pre-test to 19.5% and 22.4% in the post-test and follow-up, respectively.

**Satisfaction questionnaire results**

The majority of participants was satisfied from the learning method, the adequacy of the contents and the e-learning platform functioning. It emerges a high overall liking on the course, considering that the values are always between 4 and 5, where 5 express the highest degree of satisfaction.

**Discussion**

The aim of this study was to analyze the effectiveness of a distance learning course on genetics and genomics for Italian physicians. In recent years, a growing interest in promoting courses on genetics/genomics topics is clearly emerging [16, 17], due to rapid developments in genomic technologies and to the not sufficient knowledge of healthcare practitioners in this field [18–20]. Previous research in learning field
suggests that different aspects of an educational intervention may have impacts on its effectiveness, including the type of intervention, and the amount of practice-reinforcing strategies it contains [15]. Indeed, it appears that interactive learning, including case studies, is generally more effective at improving medical knowledge than learning based on theoretical principles alone [21].

This course represents the second Italian experience in distance training in genomics [22].

The main innovative aspect of the "Genetics and Genomics practice" course is related to the teaching methodology, oriented to an active training. The PBL methodology encourages the participants to "learn to learn" by solving real-world problems that reflect their work context [11, 12]. Schmidt et al. [10] indicated that in PBL the presentation of a problem activates the participants’ prior knowledge, enabling more effective learning to take place. Compared to a conventional approach, in PBL learning occurs in a more active way, since participants attempt to solve a problem and to identify their specific learning objectives. This way, learners face a cognitive conflict and construct their learning on their previous knowledge and experience [23]. Although in some studies CBL is contrasted to PBL in terms of structure, being guided learning, we integrated these two approaches, in order to provide a comprehensive andragogic and active orientation to the course [24]. CBL, indeed, encourages participants to integrate their learning in the context of realistic clinical environment and to connect theory to clinical practice. The learning theories applied to CBL derive mainly from adult-learning and inquiry-based learning approaches, relaying also to cognitive and social constructivist models [25].

The results of our “Genetics and Genomics practice” course suggest that distance-learning training in genetic/genomic practices represents an effective and
satisfactory method to improve physicians’ knowledge across all age groups of participants.

Among the 1637 participants who completed the course, the most represented age class was those of 51–65 years. This result may be related to the educational need of the over-50 age physicians in an innovative field as omics sciences are. In fact, most healthcare professionals did not receive an adequate training on this topic, as demonstrated by a negative correlation between time from medicine degree and omics sciences knowledge [26].

Participants declared their previous training experience on the course issues at the beginning of the course and results reports that the majority of participants had never attended other courses on that topic before.

The effectiveness of the course was measured through a Knowledge test made of a set of 10 MCQs that was repeated before the start and at the end of the course. The overall results suggest that the course improved the general level of knowledge. Nevertheless, as revealed by the stratified analysis, the improvement was not homogeneous for all the medical specialties. For example, the knowledge improvement was greater for Primary Care physicians and Sport Medicine physicians. These results accomplish our expectations regarding the course, since it was intended mainly as directed to GPs and FPs (primary care), that don’t receive a specific education in genetics during their specialization, but deal with genetic problems during the daily practice. The low pre-test score of this specialty group confirmed the educational need we hypothesized in planning the course. As for Sport Medicine physicians, the lowest pre-test score they reported could be explained with the fact that they deal with genetics more rarely than other specialties physicians; however, the great improvement obtained in the post-test
score could demonstrate the effectiveness of the course in filling the knowledge gap. On the opposite, the Gynecologists and Occupational Medicine physicians reported the lowest difference between the pre and post-test. For the first category, this might be related to the high pre-test score they reported, while for the latest both the pre-test and the post-test scores were low, if compared to the overall scores. The highest pre-test and post-test scores were registered for the Geneticists, demonstrating their pre-existing knowledge on the topic. Our results are consistent with the results reported by Michelazzo et al. [22], that analyzed the effect of a previous course in genetics and genomics for physicians, organized with different educational methodologies.

To our knowledge, this is the first study that attempts to measure the knowledge retain and the educational effects of the course after a follow-up period. This was obtained by inviting all participants to complete the same pre/post-test after a follow-up period of eight months. Although the proportion of respondents was low compared to the high number of participants, the overall scores shows that after eight months the knowledge level decreased if compared to the post-test score, but it was higher than the pre-test score, thus supporting the effectiveness in knowledge retain.

The follow-up data also allows some considerations on the self-perceived sense of competence, before and after the course, in giving information on genetic tests to patients by medical staff. Among the 258 Responders the sense of competence has improved at follow up and there was an increase in the number of doctors who felt more capable of providing information about diagnostic/prognostic utility of predictive genetic test.

Our study presents some limitations. Firstly, the effectiveness of the course could
be overestimated, due to the fact that only data of those who completed the entire course were collected. Therefore, it might be possible that the “dropouts” would have reported lower improvements or less satisfaction than the “completers”.

Secondly, the sample size of those who completed the course was quite heterogeneous in terms of specialties, not allowing a significant representation of all the discipline categories, many of which were grouped into one “other specialties” category.

Despite these limitations, the results of our study confirmed the effectiveness of genetic and genomic courses in improving participants’ literacy on omics sciences, not only in terms of knowledge, but also in terms of managing genetic information in daily practice. In particular, our results suggest that especially primary care physicians are those who can have the most important benefit from a course about this topic.

Conclusions

In conclusions, in our experience a distance-learning training in genetic/genomic practices that adopts a PBL and CBL approach was highly effective method to improve physicians’ knowledge and skills.

Declarations

**Ethics approval and consent to participate**

According to institutional regulation in Italy, ethics committee approval was not required for this study: participants were informed about methods and aims of the study and, by registering for the course on the online platform, agreed to the use of anonymous data in accordance with Italian Data Protection Regulations (Legislative
Decree no. 196/2003 amended by the decree adapting the national legal system to the GDPR 2016/679).

**Consent for publication**

Not applicable

**Availability of data and materials**

The datasets generated and analysed during the current study are not publicly available, as personal information of the participants of the course are stored, but are available from the corresponding author on reasonable request.

**Competing interests**

The authors declare that they have no competing interests.

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**Authors’ contributions**

SB and WR conceived the study and GEC, AT, AM, AF participated in its design. SB, GEC and AM developed the content of the distance learning course. AM, PC, DB, ADP realised the course in the online platform and collected the participants’ data. AT, GEC, AM, PC, DB and ADP statistically analysed data. SB, GEC, AT, AM, PC, DB and ADP critically discussed and interpreted the results of the knowledge tests.

All authors drafted and critically reviewed this manuscript and approved the final version.
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Abbreviations

PBL: Problem-based Learning
CBL: Case-based Learning
LMS: Learning Management System
GPs: General Practitioners
FPs: Family Pediatricians
CME: Continuous Medical Education
NHS: National Health Service
SQ: satisfaction questionnaire

References

1. Schneider MV, Orchard S. Omics technologies, data and bioinformatics principles. Methods Mol Biol. 2011; 719:3-30
2. Anderson MW, Schrijver I. Next Generation DNA Sequencing and the Future of Genomic Medicine. Genes. 2010; 1, 38-69
3. Muntoni F, Cross JH. Paediatric neurology: from molecular mechanisms to targeted treatments. Lancet Neurol. 2015; 14:16-8
4. Boccia S, Ricciardi W, McKee M, Federici A, Guerra R. Chief Medical Officers meeting on implementing a public health genomics approach. Epidemiol Biostat Public Health. 2015; 12:3
5. Mazzucco W, Pastorino R, Lagerberg T, Colotto M, d’Andrea E, Marotta C et al. Current state of genomic policies in healthcare among EU member states: results of a survey of chief medical officers. Eur J Pub Health. 2017;27(5):931-7.

6. Conferenza Stato-Regioni del 13.03.2013: Intesa tra il Governo, le Regioni e le Province autonome di Trento e di Bolzano sul documento recante: “Linee di indirizzo su la genomica in sanità pubblica”. (SALUTE) Codice sito: 4.10/2013/32 (Servizio III) Intesa ai sensi dell’articolo 8, comma 6, della legge 5 giugno 2003, n. 131. http://www.regioni.it/sanita/2013/03/25/conferenza-stato-regioni-del-13-03-2013-intesa-tra-il-governo-le-regioni-e-le-province-autonome-di-trento-e-di-bolzano-sul-documento-recante-linee-di-indirizzo-su-la-genomica-in-sanita-pubblica-290494. Accessed 1 Dec 2019.

7. Boccia S, Federici A, Colotto M, Villari P. Implementation of Italian guidelines on public health genomics in Italy: a challenging policy of the NHS. Epidemiol Prev. 2014 Nov-Dec;38(6 Suppl 2):29-34.

8. Boccia S, Federici A, Siliquini R, Calabrò GE, Ricciardi W on behalf of the Expert table of the Ministry of Health. Implementation of genomic policies in Italy: the new National Plan for innovation of the Health System based on omics sciences. Epidemiol Biostat Public Health. 2017; 14(4)

9. Calabrò GE, Tognetto A, Mazzaccara A, Barbina D, Carbone P, Guerrera D, Federici A, Ricciardi W, Boccia S. Omic sciences and capacity building of health professionals: a distance learning training course for Italian physicians. Ig Sanita Pubbl. 2019 Mar-Apr;75(2):105-124.

10. Schmidt HG, Rotgans JI, Yew EHJ. The process of problem-based learning: what works and why. Medical Education 2011; 45: 792-806.
11. Mazzaccara A, Barbina D, Guerrera D. Problem-based learning in distance training for health professionals: a high interactivity model. Atti Conference Mediterranean MoodleMoot, 2nd and 3rd October 2013 - Sousse, Tunisia.

12. Colucci A, Luzi AM, Fanales Belasio E, Barbina D, Mazzaccara A, Farchi S et al. A blended training programme for healthcare professionals aimed at strengthening territorial networks for the prevention and contrast of gender-based violence. Epidemiol Prev 2019; 43 (2-3):177-184.

13. McLean SF. Case-Based Learning and its Application in Medical and Health-Care Fields: A Review of Worldwide Literature. J Med Educ Curric Dev. 2016; 27:3.

14. Tognetto A, Michelazzo MB, Ricciardi W et al. Core competencies in genetics for healthcare professionals: results from a literature review and a Delphi method. BMC Med Educ 19, 19 (2019) doi:10.1186/s12909-019-1456-7

15. Pastorino R, Calabrò GE, Lagerberg T, Michelazzo MB, Boccia S. Effectiveness of educational intervention types to improve genomic competency in non-geneticist medical doctors: a systematic review of the literature. Epidemiol Biostat Public Health. 2018. 15(1)

16. Kohane I. Ten things we have to do to achieve precision medicine. Science. 2015;349(6243):37–8. https://doi.org/10.1126/science.aab1328

17. Cornel M. Evidence-Based Genetic Education of Non-Genetic-Expert Physicians: Experiences Over Three Decades in Amsterdam. Frontiers in Genetics. 2019;10.

18. Feero WG, Green ED. Genomics education for health care professionals in the 21st century. JAMA. 2011;306(9):989-90.

19. Boccia S. Why is personalized medicine relevant to public health? Eur J Public Health. 2014;24(3):349-50.

20. Hall A, Luheshi L. Signatories: Prof. Bartha Maria Knoppers, Dr Eric Meslin,
Prof. Walter Ricciardi, Dr Ron Zimmern. Personalised healthcare: bringing the future into focus. PHG Foundation. Final report of the Ickworth meeting, 2017. http://www.phgfoundation.org/documents/REPORT%20-%20Personalised%20healthcare%20bringing%20the%20future%20into%20focus.pdf. Accessed 1 Dec 2019.

21. D’Alessandro DM, Lewis TE, D’Alessandro MP. A pediatric digital storytelling system for third year medical students: the virtual pediatric patients. BMC Med Educ. 2004 Jul 19;4(1):10.

22. Michelazzo MB, Roberta Pastorino R, Mazzucco W, Boccia S. Distance learning training in genetics and genomics testing for Italian health professionals: results of a pre and post-test evaluation. Epidemiol Biostat Public Health. 2015; 12(3):311516-1/6

23. Masek A, Yamin S. A Comparative Study of the Effect of Problem Based Learning and Traditional Learning Approaches on Students' Knowledge Acquisition. International Journal of Engineering Education 2012, 28:1161-1167.

24. Thistlethwaite JE, Davies D, Ekeocha S, Kidd JM, MacDougall C, Matthews P et al. The effectiveness of case-based learning in health professional education. A BEME systematic review: BEME Guide No. 23. Med Teach. 2012; 34(6):421-444.

25. Mayo JA. Using case-based instruction to bridge the gap between theory and practice in psychology of adjustment. J Constr Psychol 2004; 17(2)137–146.

26. Hofman KJ, Tambor ES, Chase GA, Geller G, Faden RR, Holtzman NA. Physicians’ knowledge of genetics and genetic tests. Acad Med. 1993;68(8):625–32.
# Tables

## Table 1. Objectives and content of the distance learning course "Genetics and Genomics practice".

| General objective | Training of medical professionals (in particular primary care physicians) in the responsible use of "omics" technologies. |
|-------------------|---------------------------------------------------------------------------------------------------------------|
| **Specific objectives** | 1. Identify the basic concepts of human genetics  |
| | 2. Describe the main genetic / genomic tests currently available and the application |
| | 3. Describe the main applications of pharmacogenetic tests |
| | 4. Describe the main applications of genetic / genomic tests in oncology |
| | 5. Consciously manage clinical information, family history and genetic tests for optimal patient management (including possible specialist referral) |

| Topics | Case Studies |
|--------|--------------|
| **Public Health Genomics** | |
| Genetic tests in the clinical practice | 1. Pulmonary disease, sinusitis, digital hippocratism (Atypical Cystic Fibrosis) |
| | 2. Unilateral maculopathy and predictive tests (example of predictive testing directly at consumers) |
| | 3. Monitoring of pregnancy with "super-villocentesis" or "super-amniocentesis" |
| **Pharmacogenetics** | 4. Hypersensitivity to warfarin Patient with insufficient response to antiplatelet therapy Abacavir hypersensitivity syndrome |
| **Oncology genomics** | 1. Hereditary breast cancer |
| | 2. Family history of multiple cancers |
| | 3. Hereditary colon cancer |
| **Integration of genetic tests into cancer screening programs** | |

*Notes: GPs: General Practitioners; FPs: Family Pediatricians*
Table 2. Characteristics of the participants who completed the course (N=1637) and of the Responders at follow-up (N=268)
|                  | Completers   | Responders at follow |
|------------------|--------------|----------------------|
|                  | N=1637       | N=268                |
| Gender           |              |                      |
| Male             | 790 (48.3)   | 145 (54.1)           |
| Female           | 847 (51.7)   | 123 (45.9)           |
| Age (years)      |              |                      |
| <36              | 111 (6.7)    | 11 (4.1)             |
| 36-50            | 458 (28.0)   | 63 (23.5)            |
| 51-65            | 954 (58.3)   | 171 (63.8)           |
| >65              | 114 (7.0)    | 23 (8.6)             |
| Italian Region area |          |                      |
| North            | 594 (36.3)   | 101 (37.7)           |
| Center           | 413 (25.2)   | 67 (25)              |
| South            | 630 (38.5)   | 100 (37.3)           |
| Medical specialty |          |                      |
| Primary care     | 340 (20.8)   | 63 (23.5)            |
| GPs              | 226 (13.8)   | 35 (13.0)            |
| FPs              | 93 (5.7)     | 23 (8.6)             |
| Continuity of care Physicians | 21 (1.3) | 5 (1.9) |
| Hygiene &Public Health | 190 (11.6) | 40 (14.9) |
| Pediatrics       | 174 (10.6)   | 29 (10.8)            |
| Psychiatry       | 118 (7.2)    | 19 (7.1)             |
| Sports Medicine  | 68 (4.2)     | 12 (4.5)             |
| Genetics         | 60 (3.7)     | 1 (0.4)              |
| Occupational Health Medicine | 54 (3.3) | 12 (4.5) |
| Gynecology and obstetrics | 53 (3.2) | 3 (1.12) |
| Other specialties | 580 (35.4)  | 95 (35.5)            |
| Professional status |        |                      |
| Private health facilities/NHS Employees | 913 (55.8) | 148 (55.2) |
| Freelancers      | 337 (20.6)   | 49 (18.3)            |
| Private contractors with NHS | 359 (21.9) | 64 (23.9) |
| Without occupation | 28 (1.7)   | 7 (2.6)              |
Table 3. Knowledge level of the Completers before and after the course (N=1637).

| Nº | Question                                                                                                                                                                           | Correct answers before the course (T0) |
|----|-----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------|
| 1  | Which of the following groups of diseases is characterized by the interaction between genes and the environment?                                                                | 917 (56.0)                             |
| 2  | What is the name of the study of DNA polymorphisms in order to predict the safety and efficacy of drugs?                                                                     | 1319 (80.6)                            |
| 3  | Is the evaluation of the hereditary-family risk of a tumor carried out as part of the oncological screening pathways?                                                              | 515 (31.5)                             |
| 4  | What is a predictive test?                                                                                                                                                         | 1261 (77.0)                            |
| 5  | What are the main models of Mendelian heritage?                                                                                                                                   | 1446 (88.3)                            |
| 6  | What do pharmacogenetic tests predict?                                                                                                                                           | 1263 (77.2)                            |
| 7  | What coverage does participation in cancer screening programs for breast and colorectal cancer reach in our country?                                                             | 336 (20.5)                             |
| 8  | In hereditary forms of cancer, what is the transmission of the gene involved?                                                                                                     | 705 (43.1)                             |
| 9  | What types of analysis are performed with Next Generation Sequencing (NGS) techniques?                                                                                            | 678 (41.4)                             |
| 10 | What is the role of diagnostic genetic tests in the field of hereditary tumors?                                                                                                     | 1294 (79.1)                            |

Table 4. Completers’ pre-test versus post-test average scores according to several participants’ characteristics.
| Overall (N=1637) | Pre-test (T0) mean score | Post-test (T1) mean score |
|------------------|--------------------------|--------------------------|
|                  | 59.46                    | 71.42                    |

| Gender           |                           |                           |
|------------------|--------------------------|--------------------------|
| Male (N= 790)    | 60.28                    | 72.77                    |
| Female (N= 847)  | 58.70                    | 70.17                    |

| Age              |                           |                           |
|------------------|--------------------------|--------------------------|
| <56 years (N= 814) | 61.81                    | 72.33                    |
| ≥56 years (N= 823) | 57.14                    | 70.52                    |

| Region           |                           |                           |
|------------------|--------------------------|--------------------------|
| North (N= 594)   | 60.24                    | 73.38                    |
| Center (N= 413)  | 59.54                    | 71.94                    |
| South (N= 630)   | 58.68                    | 69.24                    |

| Medical specialty |                           |                           |
|-------------------|--------------------------|--------------------------|
| Primary care (N= 340) | 56.97                    | 71.29                    |
| Hygiene & Public Health (N= 190) | 60.21                    | 70.63                    |
| Pediatrics (N= 174) | 62.59                    | 73.68                    |
| Psychiatry (N= 118) | 57.97                    | 70.68                    |
| Sport Medicine (N= 68) | 53.09                    | 68.38                    |
| Genetics (N= 60)  | 72.83                    | 83.5                     |
| Occupational Health Medicine (N= 54) | 58.89                    | 68.33                    |
| Gynecology and obstetrics (N= 53) | 63.40                    | 72.26                    |
| Other specialties (N= 580) | 59.10                    | 70.55                    |

| Professional status |                           |                           |
|---------------------|--------------------------|--------------------------|
| Private health facilities/NHS Employees (N= 913) | 59.55                    | 71.04                    |
| Freelancers (N= 337) | 60.68                    | 71.39                    |
| Private contractors with the NHS (N= 359) | 57.86                    | 72.23                    |
| Without occupation (N= 28) | 62.5                     | 73.93                    |

Table 5. Knowledge level of the Responders before, after the course and at follow-up (N=268)
| N° | Question                                                                 | Correct answers (T N (%) |  |
|----|--------------------------------------------------------------------------|--------------------------|---|
| 1  | Which of the following groups of diseases is characterized by the interaction between genes and the environment? | 150 (56.0)               |  |
| 2  | What is the name of the study of DNA polymorphisms in order to predict the safety and efficacy of drugs? | 210 (78.4)               |  |
| 3  | Is the evaluation of the hereditary-family risk of a tumor carried out as part of the oncological screening pathways? | 64 (23.9)                |  |
| 4  | What is a predictive test?                                               | 217 (81.0)               |  |
| 5  | What are the main models of Mendelian heritage?                          | 243 (90.7)               |  |
| 6  | What do pharmacogenetic tests predict?                                   | 210 (78.4)               |  |
| 7  | What coverage does participation in cancer screening programs for breast and colorectal cancer reach in our country? | 53 (19.8)                |  |
| 8  | In hereditary forms of cancer, what is the transmission of the gene involved? | 119 (44.4)               |  |
| 9  | What types of analysis are performed with Next Generation Sequencing (NGS) techniques? | 100 (37.3)               |  |
| 10 | What is the role of diagnostic genetic tests in the field of hereditary tumors? | 204 (76.1)               |  |

Table 6. Responders’ answers on the perceived competence about the capability to meet the patients request on genetic tests (N=258)
When asked by patients, I was able to provide information about diagnostic/prognostic utility of predictive genetic test

|                | Before the course (T0) * N (%) | After the course (T1) N (%) |
|----------------|-------------------------------|-----------------------------|
| Never          | 22 (8.5)                      | 31 (12:1)                   |
| Rarely         | 38 (14.7)                     | 30 (11:7)                   |
| Occasionally   | 20 (7.7)                      | 18 (7.0)                    |
| Sometimes      | 37 (14.3)                     | 26 (10:2)                   |
| Regularly      | 22 (8.5)                      | 21 (8:2)                    |
| Usually        | 17 (6.6)                      | 18 (7:0)                    |
| Always         | 8 (3.1)                       | 11 (4:3)                    |
| Not applicable | 94 (36.4)                     | 101 (39:6)                  |

* Referred to 12 months before the course’s start

Figures
Figure 1

Flowchart of cohort selection of participants. Note: CME: Continuous Medical Educ