

# Addressing the gap between genetics knowledge and clinical practice: a pilot study to implement genetics education among physicians in Italy

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As a result of large investments in basic science, the genomic discoveries have brought outstanding advances in understanding the molecular basis of human health. Hundreds of genes whose variations contribute to human diseases, or patients' responses to drug treatments or even to vaccination have been discovered, laying the foundation for a paradigm shift in healthcare (1). This new healthcare model, commonly known as personalised medicine, has wide ranging implications for all stakeholders, from individual European citizens to physician practice, to the institutional, regional and European policymakers (2). The challenge is to deliver the benefits of this work to patients, and education of healthcare professionals is likely to play an important role in bringing this about, as they will be called upon to make decisions based on complex biological, environmental and lifestyle information (3). Needs for genomics understanding among physicians, citizens and policy makers have been widely documented in the literature. We have evidences that most of the physicians recognize their knowledge deficits and are interested in specific training (4, 5).

With the introduction of "omics" data as a common component of patients' health records, physicians will need a clear understanding of the principles underlying their interpretation. They will need to be able to critically review the information provided to them and that will require a solid grounding in genetics, biological methodology and statistical interpretation. Consequently, educational efforts to promote understanding of patients' data are essential for the implementation of personalised medicine.

Beside this, it is widely documented that the general public interest on genetic tests is rapidly increasing, so that citizens understanding and perception on pros and cons of personalized medicine should be adequately sustained (6, 7). And again, it is evident that policy makers need to be aware of genomics implications in order to implement adequate policies and legislation related to a more complex scenario in term of genomics innovations, ethical, legal and social issues and management approach.

Indeed, inappropriate prescribing of genomic tests for complex diseases, mainly referred both to a prescribers lack of knowledge and to an increased requests by the citizens, generates some effects of public health concern as the "cascade effect" (8), consisting in a series of not evidence-based diagnostic investigations, resulting from a positive genomic test. The proliferation of small/medium size laboratories, offering genetic tests without the guarantee of minimum quality standards, or without providing appropriate genetic counseling, is also an issue in the health care provision.

How above is in contrast with any principle of sustainability for a public National Health System, and this is even more evident in light of the current economic crisis affecting the Western Countries, prioritizing

austerity measures and health expenditures containment and making any initiative towards a systematic assessment of the genome based tests an investment for the future. Lastly, the availability of direct to consumer (DTC) genetic testing (GT) for health-related purposes is also rapidly increasing worldwide, expressing an urgent need of regulation of this phenomenon (9, 10).

In this context significant efforts have been made by the Italian Health Ministry with the forthcoming publication of a specific action plan on genomics and predictive medicine within the 2010-2012 National Prevention Plan (NPP) (11). The PHG action plan aims to address more in depth how to translate the genomics knowledge into public health for the benefits of population health, and it has been drafted with the scientific support of the Institute of Hygiene of Università Cattolica del Sacro Cuore in close collaboration with the GENISAP Network (12-14). The PHG action plan focus on five main strategic objectives, including: the definition of areas of intervention of PHG; the collection of evidence on current PHG experience in the country; the definition of the best instruments for the promotion of genome-based knowledge among health professionals and citizens; the development of evidence-based recommendations for the appropriate use of genetic testing of complex diseases; the identification of the critical points for the translation of genomics into clinical practice. The fulfilment of all these objectives is granted by three major mainstays: the systematic health technology assessment of genetic tests for complex diseases currently in use and a pre-marketing assessment of those not yet currently available; the promotion of genomics education towards physicians (general practitioners first) and capacity building for the potential stakeholders in the healthcare provision and management; the promotion of basic genomic health literacy for general population, in order to raise awareness of potential benefits, limits and risks of genomic technologies (15).

In Europe a solid background has been experienced in the professionals' educational and training fields: within EuroGentest, a project funded by the European Commission to harmonize the process of genetic testing across Europe, the suggested core competences in genetics for health professionals, generalists or specialising in a field other than genetics, have been defined (16).

An innovative educational and training pilot programme is currently undergoing in Italy, thanks to a grant of the National Centre for Disease Control (Centro per il Controllo delle Malattie, CCM) of the Italian Health Ministry. A panel of experts, representative of six Italian regions (Emilia-Romagna, Latium, Liguria, Lombardy, Tuscany, Veneto), has defined the contents of a modular course to be primarily administered through a virtual e-learning platform to a population of genomic tests potential prescribers, recruited with the support of the scientific societies. Distance learning through the web (known under the acronym of FAD in Italy) was adopted in reason of being an educational strategy that allows the learner to participate in a structured set of learning activities to encourage independent and personalized learning mode, discontinuous in time and space (17). The aim of the training programme is to support general practitioners, public health physician, neurologists, gynaecologists, oncologists on the appropriate prescription of genomic testing by addressing some key issues. Innovation in contents is represented by an interdisciplinary approach to the matter. Public health specialists, geneticists, clinical specialists, and general practitioners have developed together each didactic module, ranging from basics of medical genetics and genetic testing framework, to genetic testing pathways, including also specific topics on genetic testing ethical, legal and social implications, medical genetics service and genetic counselling. Practical exercises are also reported, in order to stimulate GPs and specialists on "real world" situations they might face in their practice. The programme implementation is currently underway and, in case of a positive evaluation by the Ministry of Health, the pilot project will be likely extended to the whole Country.

In conclusion, we truly believe that implementation of education and training programmes dedicated to genomic testing prescribers will lead to an appropriate use of genomic knowledge and technologies. We hope that the Italian experience might be of interest for the other high-income countries, contributing for a shift in culture regarding prescription and appropriate use of predictive genomic tests for complex diseases.

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