

## Challenges for the Italian Public Health Genomics Task Force

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Dear Sirs,

In recent years, the results of the Human Genome Project have led to huge expectations relating to the rapid implementation of its results in the field of prevention. The possibility to provide early detection for those individuals more susceptible to complex diseases, because of their genetic *make-up*, might theoretically result in individualized primary (e.g., chemoprevention) and secondary prevention (e.g., assiduous monitoring) programs.

The issue of an individual's inherited susceptibility to develop complex diseases, however, is much more complicated than it first appears, because of the delicate links between scientific developments, technical applications and ethical considerations. Consequently, besides the framework required for the critical evaluation of genetic testing (analytical validity, clinical validity and clinical utility of the test), we must also consider these three fundamental aspects:

- From pooled and meta-analyses of genetic association studies, it appears that most of the commonly identified genetic variants (polymorphisms) confers a low risk of disease (on average ORs of 1.5, if not lower), which only increases with the interaction of well-known environmental risk factors
- From preliminary studies, the result from a 'positive' genetic testing does not seem to motivate behavioural changes
- There is a need for an evidence-based approach to genetic testing that integrates the cost-utility evaluation of each available genetic test [Health Technology Assessment (HTA) framework] while considering the weight of the relevant ethical issues.

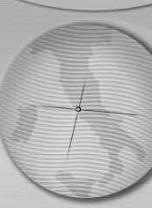
With these considerations in mind, on 10<sup>th</sup> July 2006 the kick-off meeting of the first Italian Task Force on Public Health Genomics (ITF-PHG) took place at the Institute of Hygiene in the Faculty of Medicine of the Catholic University of the Sacred Heart (UCSC) in Rome, coordinated by Walter Ricciardi (Director of the Institute of Hygiene-UCSC) and Stefania Boccia.

The kick-off meeting, as well as the second one held on 5<sup>th</sup> October, 2006, focused on the necessity to integrate Genetic and Public Health practice with policy in Italy. Previously there has been limited collaboration between the two disciplines which is characterized by a subsequent lack of understanding on both sides. Participation within the group is varied and includes six university academics from the fields of PH and HTA, five from the Human Genetic field, and one from social sciences.

Our first step was to disseminate information regarding Public Health Genomics knowledge inside the Italian Scientific Society of Public Health and Human Genetics. This was undertaken during the months of October and November 2006, by taking advantage of poster opportunities at both of the Annual National Conferences. Furthermore, it was suggested to the Presidents of the Italian Society of Public Health and Human Genetics, that working groups and teaching Courses within the two societies should be established in time for the next National Conferences in 2007.

Secondly, we discussed funding opportunities. In the National Plan for Research 2005, the former Italian Minister reported on genetic epidemiology and the necessity for it to be funded as field of research in the forthcoming years. However the term 'Public Health Genomics' was not yet widely recognised, and the general impression from the participants of the meeting was that PHG did not exist in Italy, thus highlighting an important issue for our group to address.

The main outcome of the second meeting was the plan to implement an evidence-based model for the evaluation of the appropriateness of genetic testing supplied in Italy, in view of the growing costs to the National Health Service. In this regard, the ITF-PHG created an action plan to be implemented, which included the wide dissemination of a document on the subject of PHG research in Italy (covering basic concepts and ethical, law and social aspects from an Italian perspective, and an inventory of the key experts



in both fields), as well as the development of a specific project to seek funding from both the Italian Centre for Disease Control and Prevention (CCM) and the Italian Ministry of University and Research.

In conclusion the group agreed to raise awareness within the Italian scientific community about the importance of PHG as a field of research, and a press release concerning both meetings was sent to the University Press Office which was then further disseminated on the newspaper "Il Sole 24 Ore Sanità" (14/11/2006).

**ITF-PHG participant list:**

Prof. Walter Ricciardi (Director Institute of Hygiene, UCSC, Rome), Prof. Elisa Calzolari (Director of the Institute of Human Genetics, University of Ferrara; member of the Board of the Italian Society of Human Genetics), Prof Silvio De Flora (Director Hygiene Section, Department of Health Sciences, University of Genova), Prof. Alberto Izzotti (Hygiene Section, Department of Health Sciences, University of Genova), Prof. Paolo Mastroiacovo (Alessandra Lisi International Centre on Birth Defects, Director), Prof. Giovanni Neri (Director of the Institute of Human Genetics, UCSC, Rome), Prof. Paolo Villari (Hygiene Section, Department of Experimental Medicine and Pathology, University of Rome "La Sapienza"), Dr. Stefania Boccia (Head of the Genetic Epidemiology and Molecular Biology Unit, Institute of Hygiene, UCSC, Rome), Dr. Anna Baroncini (Head of the Service of Medical Genetics-ASL, Imola), Dr. Franca Dagna Bicarello (President of the Society of Human Genetics, Head of the Laboratory of Human Genetics, Hospital "Galliera", Genova), Dr. Domenico Coviello (Head of the Laboratory of Human Genetics, Hospital "Maggiore Policlinico, Mangiagalli e Regina Elena-IRCCS", Milan), and Dr. Laura Murianni (National Observatory on Health in the Italian Regions, Institute of Hygiene, UCSC).